

ORIGINAL RESEARCH

Efficiency of refractive error improvement for people with oculocutaneous albinism in India

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ABSTRACTS

Introduction: Albinism originates from the Latin word albus, which means white. It refers to a collection of inherited illnesses where the production of the pigment melanin is either absent or decreased. Oculocutaneous albinism (OCA) is a diverse and inherited illness that results in a loss of colour in the skin, hair, and eyes. It is also associated with visual problems such sensitivity to light, misalignment of the eyes, impaired vision, and involuntary eye movements. **Materials and methods:** This study was conducted in a tertiary care hospital in West Bengal. It was a descriptive community-based study using a cross-sectional design. The ophthalmology department gathered 50 individuals who agreed to enrol. The diagnostic criteria for OCA were the existence of iris transillumination, retinal hypopigmentation, and depigmentation of the skin, hair, and nails. **Results:** 100 individuals with OCA were included, with an average age of 20 years. Of these, 60 (60%) were male and 40 (40%) were female. The highest percentage of participants (40%) fell within the age range of 16 to 25 years. According to the categorization of the World Health Organisation, based on the best corrected distance visual acuity, 8% of 100 eyes had normal vision, 70% had moderate visual impairment, 15% had severe visual impairment, and 7% were blind. **Conclusion:** Albinos have a significant occurrence of refractive, non-refractive, and mixed eye diseases. People with albinism experienced noticeable improvement in their ability to see and function after receiving optical correction and alignment, even when their overall visual acuity was below average. It is advisable to promote refractive correction for those with albinism.

Keywords:

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INTRODUCTION

Albinism is derived from the Latin word albus, which means white. It refers to a collection of inherited conditions where the production of the pigment melanin is either missing or decreased. Currently, it is categorised based on the gene that is impacted, rather than being classed as partial or entire, tyrosinase positive or tyrosinase negative.¹ Albinism is a diverse range of hereditary illnesses that impact 1 in 20,000 people globally, however the occurrence of the many kinds of albinism differs significantly among different ethnicities. It is the result of a lack of pigmentation, and is clinically classified as either ocular or oculocutaneous albinism.²⁻⁴

Oculocutaneous albinism (OCA) is a diverse and inherited illness that results in a deficiency of colour in the skin, hair, and eyes. It is also associated with visual impairments including sensitivity to light,

misalignment of the eyes, reduced vision, and involuntary eye movements.⁵⁻⁶ The classification of albinism based on physical characteristics is either oculocutaneous albinism (OCA) or ocular albinism (OA). OCA individuals have decreased melanin in their eyes, skin, and hair, while OA only affects the eyes with diminished or missing melanin.⁷

OCA exhibits notable optical imperfections, such as a considerable corneal astigmatism, foveal hypoplasia, and aberrant crossing of optic nerve fibres. The atypical crossing is believed to define the precise target of neurons and the incorrect routing of retinogeniculate projections, leading to strabismus and decreased depth perception.⁸⁻¹⁰ As a result, visual acuity (VA) is typically decreased, resulting in vision problems, and individuals with this condition often have intense sensitivity to light (photophobia). Significant refractive defects, including as

astigmatism, are common.¹¹ The majority of the population resides in rural areas that are cut off from healthcare facilities and have limited understanding of medical concerns. Individuals with albinism, as a result, are frequently not well comprehended and find themselves entangled in a realm of spiritual convictions and superstition. People who seek help rarely get glasses because many specialists in rural North India assume that they have little potential for improving their vision.¹²

MATERIALS AND METHODS

This study was conducted in a tertiary care hospital in West Bengal. It was a descriptive community-based study using a cross-sectional design. The ophthalmology department gathered 50 individuals who agreed to enrol. The diagnostic criteria for OCA were the existence of iris transillumination, retinal hypopigmentation, and depigmentation of the skin, hair, and nails. Among the 50 participants, 5 were already wearing glasses. They were reassessed and given new glasses with lenses that change colour in response to light.

A record was made of the demographic profile, history of chief complaints, previous use of low vision equipment, and usage of any refractive correction. Unassisted and assisted visual acuity (VA) was assessed using log MAR charts for individuals who cannot read and those who can read, as appropriate. This was done in combination with retinoscopy and subjective refraction. Eye movement, misalignment of

the eyes, merging of visual images, and involuntary eye movements were evaluated. An examination of the front and back parts of the eye was conducted after the pupils were dilated.

Cycloplegic refraction was performed 30 minutes after applying three drops of 1% cyclopentolate, with a 5-minute interval between each drop. Individuals with normal vision were not included in the study.

Participants were interviewed via telephone three weeks after receiving their glasses. Participants or parents provided information on compliance with wearing glasses, which was categorised as excellent (>75% of awake hours), good (50–75%), fair (26–50%), or poor (<25%). The participants were given unstructured open-ended inquiries regarding the influence of eyeglasses on their overall well-being and everyday activities.

RESULTS

100 individuals with OCA were included, with an average age of 20 years. Of these, 60 (60%) were male and 40 (40%) were female. The highest percentage of participants (40%) was within the age range of 16 to 25 years (Table 1).

According to the classification of the World Health Organisation, based on the best corrected distance visual acuity, 8%, 70%, 15%, and 7% of 100 eyes had normal vision, moderate visual impairment, severe visual impairment, and blindness, respectively (Table 2).

Table 1: Distribution of participants by age and sex

Age (years)	Sex		Total	Percentage (%)
	Male	Female		
0-5	10	4	14	14
6-15	12	9	21	21
16-25	22	18	40	40
26-35	10	5	15	15
36-50	6	4	10	10
Total	60 (60%)	40 (40%)	100	100

Table 2: Distance visual acuity by eye at presentation and after correction

Distance VA	N (%)		Classification of VI
	Presenting VA	Corrected VA	
6/6-6/18	10 (5)	14 (7)	Normal
<6/18-6/60	80 (40)	140 (70)	Moderate
<6/60-3/60	60 (30)	30 (15)	visual impairment
<3/60-PL	50 (25)	16 (8)	Severe visual
Blindness Mean distance VA in logMAR	1.19±0.29	0.97±0.27	Impairment Blindness

Table 3: Near visual acuity by eye by distance; 200 eyes

Near visual acuity (meters)	N (%)	
	Presenting visual acuity	Corrected visual acuity
1	40 (20)	60 (30)
1.25	60 (30)	50 (25)
1.5	4 (2)	6 (3)
1.6	0	4 (2)
2	64 (32)	50 (25)

2.25	0	8 (4)
2.5	8 (4)	0
3.2	4 (2)	10 (5)
4	10 (5)	8 (4)
4.5	2 (1)	4 (2)
5	8 (4)	0
Total Mean VA	2.13±1.19	1.83±0.91

Table 4: Refractive error analysis

Refractive error	N (%)
Myopia	40 (20)
Hypermetropia	20 (10)
Myopic astigmatism	60 (30)
Hypermetropic astigmatism	80 (40)
Total	200 (100)

Table 5: Mean strabismus

Strabismus in mean	Without glasses	With glasses	p-value
Near (prism diopters)	17.13±19.3	13.23±17.59	0.001
Distance (prism diopters)	17.29±19.54	13.20±17.21	0.001

DISCUSSION

People with oculocutaneous albinism experience visual impairment that affects their ability to interact socially, leading to challenges in cognitive, emotional, social, and academic aspects of their lives.¹³In our research, 95% of the eyes experienced visual impairment, with the majority (70%) having moderate visual impairment and 8% being blind, which is similar to the findings of Eballe et al,¹⁴A different study conducted in Nepal found that 56% of the eyes had a moderate level of vision impairment, while 8% were classified as blind.¹³In our research, hypermetropic astigmatism was the most common condition (n=80, 40%). However, in a study conducted in Nepal by S. Khanal,¹³et al, myopic astigmatism was most common.

Out of the 44 individuals, 75% were male and 31 were females who were 25 years old or younger. These participants had tertiary education and were often students or unemployed. A comparable age distribution was noted in Europe, South Africa,¹⁵Nigeria¹⁶and Tanzania,¹⁷in research conducted with individuals with albinism. The increased inclination of younger individuals to seek medical solutions for their health or visual impairments may explain this.¹⁸The observed educational profile, in line with the age distribution of the participants, likely represents the reported average reading skills.^{19,20}and cognitive growth among individuals with OCA.²¹Nevertheless, this contradicts the conclusions made by Okoro et al.²²that nearsightedness, a frequent vision problem in albinism, is linked to cognitive difficulties. Thus, the current findings do not indicate a requirement for establishing a specialised learning environment for individuals with albinism.

In our research, half of the participants (50 patients) achieved fusion with or without glasses, whereas only

one participant achieved fusion alone with spectacles. No one had stereopsis, whether with or without glasses. A research of comparable size in the United States found that two individuals saw an improvement in vision wearing glasses, whereas one individual's vision worsened.²³

The study conducted in the US found that a greater number of patients were compliant with wearing spectacles during their follow-up visits to the outpatient clinic. Specifically, 29 patients (83%) had excellent compliance, 4 patients (11%) had acceptable compliance, and 2 patients (6%) had poor compliance. This study was conducted in an urban context where the majority of patients were located.²³Our study might have had lesser adherence since this rural group, unlike the US population, were wearing glasses for the first time and did not have an extensive follow-up programme to help with frame fitting.

The specific reason behind visual impairment in individuals with albinism is not understood, but it is believed to be related to foveal hypoplasia, nystagmus, and refractive error. In addition, amblyopia, which can occur when refractive correction is delayed, may also be a factor. This suggests that, in addition to correcting refractive errors in a timely manner, it is important to ensure that various visual aids are readily accessible to albinos to help mitigate the visual effects of these abnormalities. Some patients had other eye problems such as pterygium, pingueculum, and ptosis, which were not connected to albinism. This discovery cannot be compared to other similar studies because none of them provided data that could be compared. This highlights the importance for future researchers to recognise and effectively handle additional illnesses that may have negative effects on vision or eye health.

CONCLUSION

Albinos have a high occurrence of refractive, non-refractive, and mixed ocular diseases. To reduce the visual effects of these conditions, the researchers suggest providing eye care treatments to albinos in a timely manner, ensuring they have unrestricted access to these services, and raising awareness about their needs. People with albinism experienced noticeable improvement in their ability to see and function after receiving optical correction and alignment, even when their overall visual acuity was below normal. It is advisable to promote the use of refractive correction for individuals with albinism.

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