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Effectiveness of Refractive Error Correction for People with Oculocutaneous Albinism in Western India

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ABSTRACT

Albinism comes from the Latin albus, which meaning white, and is a group of hereditary disorders in which the biosynthesis of the pigment melanin is absent or reduced. Oculocutaneous albinism (OCA) is a heterogeneous and autosomal recessive disorder that involves a lack of pigment in the skin, hair and eyes and is accompanied by optic defects such as photophobia, strabismus, poor vision and nystagmus. This was a cross sectional descriptive community based study conducted at tertiary care hospital at Western Uttar Pradesh. The department of opthalmology collected 50 people accepting enrollment. The diagnostic criteria for OCA were presence of iris transillumination, retinal hypopigmentation and depigmentation of the skin, hair, and nails. Hundred patients with OCA were included, mean age 20 years with 60 (60%) male and 40 (40%). The largest proportions of participants (40%) were between16 and 25 years. Using world health organization classification based on best corrected distance visual acuity, 8-70%, 15-7% of 100 eyes had normal vision, moderate VI, severe VI and blindness respectively. Conclusion: There is high prevalence of refractive, non-refractive and mixed ophthalmic disorders among albinos. There was significant improvement in visual acuity and function following optical correction and alignment in people with albinism, despite overall subnormal acuity. Refractive correction should be encouraged for people with albinism.

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Key Words

Oculocutaneous albinism, visual impairment, refractive error, ophthalmic disorders

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INTRODUCTION

Albinism comes from the Latin albus, which meaning white and is a group of hereditary disorders in which the biosynthesis of the pigment melanin is absent or reduced. Currently it is classified according to the gene affected and no longer as partial or total, tyrosinase positive or tyrosinase negative[1]. Albinism is a heterogeneous group of genetic disorders that affect 1 in 20,000 individuals worldwide, although the prevalence of the different subtypes of albinism varies considerably among the different ethnic backgrounds. It is caused by deficiencies in pigmentation and clinically is divided into ocular and oculocutaneous albinism[2-4].

Oculocutaneous albinism (OCA) is a heterogeneous and autosomal recessive disorder that involves a lack of pigment in the skin, hair and eyes and is accompanied by optic defects such as photophobia, strabismus, poor vision and nystagmus[5-6]. The phenotypic classification of albinism is either oculocutaneous albinism (OCA) or ocular albinism (OA). OCA people have reduced melanin in the eyes, skin and hair, whereas OA involves reduced or absent melanin only in the eyes[7].

OCA has significant optical defects including large corneal astigmatism, foveal hypoplasia and abnormal decussation of optic nerve fibers. The abnormal decussation is thought to determine the neuronal target specificity and misrouting of retinogeniculate projections resulting in strabismus and reduced stereoscopic vision[8-10]. Consequently, VA is generally reduced, leading to visual impairment, and cases tend to have severe photophobia. High refractive errors, including astigmatism occurs frequently[11]. Most the popultion live in rural areas isolated from health care services with minimal knowledge of medical conditions. People with albinism therefore remain poorly understood often caught in a world of spiritual beliefs and superstition. Those that do seek assistance seldom receive glasses because most rural Western Indian professionals presume they have poor potential for visual improvement[12].

MATERIALS AND METHODS

This was a cross sectional descriptive community based study conducted at tertiary care hospital at Sarojini Naidu Medical College Agara. The department of opthalmology collected 1000 people accepting enrollment. The diagnostic criteria for OCA were presence of iris transillumination, retinal hypopigmentation and depigmentation of the skin, hair and nails. Out of 100 participants, 5 already had glasses. They were re-evaluated and prescribed new glasses with photochromatic lenses.

Demographic profile, history of chief complaints, previous use of low vision devices and use of any refractive correction were recorded. Unaided and aided VA was measured by using log MAR illiterate and literate charts (whichever appropriate) along with retinoscopy and subjective refraction. Extra ocular motility, strabismus, fusion and nystagmus were assessed. Anterior and posterior segment evaluation was performed under mydriasis.

Cycloplegic refraction was carried out 30 min after instillation of three drops of 1% cyclopentolate 5 min apart. Participants who had emmetropia were excluded from the study. Participants were interviewed by phone 3 weeks after glasses dispensed. Compliance with glasses wear was recorded as excellent (>75% of awake hrs), good (50-75%), fair (26-50%), or poor (<25%) according to the information given by participants or parents. They were asked unstructured open ended questions about the impact of glasses on their quality of life and activities of daily living.

RESULTS

100 patients with OCA were included, mean age 20 years with 60 (60%) male and 40 (40%). The largest proportions of participants (40%) were between16 and 25 years (Table 1). Using world health organization classification based on best corrected distance visual acuity, 8-70%, 15-7% of 100 eyes had normal vision, moderate VI, severe VI and blindness respectively (Table 2).

DISCUSSIONS

Individuals with oculocutaneous albinism have visual impairment that compromises their social interactionscompared to their peers, resulting in cognitive, emotional, social and academic difficulties[13]. In our study, 95% of the eyes had visual impairment with the majority (70%) having moderate visual impairment and 8% blind, similar to Eballe *et al*.[14], Another study in Nepal reported that 56% of the eyes were moderate visual impairment and 8% were blind[13]. In our study, hypermetropic astigmatism was most prevalent (n = 80, 40%) but in the study in Nepal by Khanal *et al*.[13] myopic astigmatism was most common.

The 75 (75%) of the participants among 44 were male and 31 were females aged 25 years or younger, who possessed tertiary education and were frequently students or unemployed. A similar age distribution was observed in Europe, South African,[15] Nigeria[16] and Tanzania,[17] in studies among albinos. The higher tendency of younger people to seek for medical solution to their health or visual disability may account for this[18] The observed educational pro le, consistent with the participant's age distribution, probably reects the reported normal reading ability[19,20] and intellectual development among persons with OCA[21] However, this challenges the ndings by Okoro *et al*.[22]

Table 1: Distribution of participants by age and sex

	Sex			
Age (years)	Male	Female	Total	Percentage
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

	N (%)		
Distance VA	Presenting VA	Corrected VA	Classification of VI
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 log MAR	1.19±0.29	0.97±0.27	Impairment Blindness

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

	N (%)	
Near visual acuity (meters)	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

that myopia, a common refractive anomaly in albinism, is associated with intellectual impairment Therefore, the present data do not support the need for creating special learning environment for albinos.

In our study, 50% (50 patients) had fusion with or without glasses and one only with glasses. None had stereopsis either with or without glasses. A similar sized study in the United States reported two individuals who gained and one who lost fusion with glasses[23]. The US study showed a higher spectacle compliance during follow up visits to the outpatient clinic with excellent in 29 patients (83%), fair in 4 (11%) and poor in 2 (6%) in an urban setting where most[23]. Our study may have lower compliance because this rural population, in contrast to the US population, were wearing glasses for the first time and had no active follow up program to assist with fitting of frames.

Although the exact cause of visual impairment in people with albinism is unknown, foveal hypoplasia, nystagmus and refractive error have been implicated. Additionally, amblyopia, resulting from delay in refractive correction, might be contributory. This implies that, beyond timely refractive correction, other visual/optical aids to alleviate the visual consequences of these abnormalities should be made widely available and accessible to albinos. Miscellaneous non-albinism-related ophthalmic disorders comprising pterygium, pingueculum and ptosis were seen in a minority of participants. This finding could not be compared with other related surveys as none reported comparable data. This underscores the need for future investigators to identify and adequately manage co-morbid miscellaneous disorders with potentially adverse visual or ocular health implications.

CONCLUSION

There is high prevalence of refractive, non-refractive and mixed ophthalmic disorders among albinos. To alleviate the visual consequences of these disorders the investigators recommend timely provision of, unrestricted access to and needs awareness creation among albinos on, appropriate eye care services. There was significant improvement in visual acuity and function following optical correction

and alignment in people with albinism, despite overall subnormal acuity. Refractive correction should be encouraged for people with albinism.

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