International Journal of Medical Ophthalmology

E-ISSN: 2663-8274
P-ISSN: 2663-8266
www.ophthalmoljournal.com
IJMO 2022; 4(1): 14-16
Received: 12-11-2021
Accepted: 21-12-2021

Ganeshkumar Jayakrishnan
Assistant Professor,
Department of Ophthalmology,
Dhanalakshmi Srinivasan Medical College and Hospital,
Siruvachur, Perambalur, Tamil Nadu, India

Nallamuthu P
Professor, Department of Ophthalmology, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India

Giridhar PS
Professor and Head of Department, Department of Ophthalmology, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India

Corresponding Author:
Ganeshkumar Jayakrishnan
Assistant Professor,
Department of Ophthalmology,
Dhanalakshmi Srinivasan Medical College and Hospital,
Siruvachur, Perambalur, Tamil Nadu, India

Effectiveness of refractive error correction for people with oculocutaneous albinism in South Indian population

Ganeshkumar Jayakrishnan, Nallamuthu P and Giridhar PS

DOI: https://doi.org/10.33545/26638266.2022.v4.i1a.107

Abstract
Introduction: Albinism comes from 'albus', the Latin word meaning white, and refers to a group of hereditary disorders wherein 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 associated with ocular and visual defects such as photophobia, strabismus, nystagmus and low vision.
Materials and Methods: This was a cross sectional descriptive community based study conducted at a tertiary care medical college hospital. The Department of Ophthalmology enrolled 39 subjects satisfying the inclusion criteria. The diagnostic criteria for OCA patients included in the study, were those having ocular features of iris transillumination and retinal hypopigmentation in addition to depigmentation of the skin, hair and nails.
Results: 39 patients with OCA were included, mean age 20 years. The largest proportion of participants (40%) included those between 16 and 25yrs. Using World Health Organization classification based on best corrected distance visual acuity, 8%, 70%, 15% and 7% of 78 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.

Keywords: Albinism, melanin, low vision, refractive error

Introduction
Albinism comes from 'albus', the Latin word meaning white, and refers to a group of hereditary disorders wherein 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 comprises of symptom complex defects 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 ocular & visual 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). Those afflicted with OCA have reduced melanin involving the eyes, skin and hair, whereas OA tend to have reduced or absent melanin only in the eyes [7]. OCA has significant ocular 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]. In addition, high refractive errors including astigmatism occurs frequently [11]. Consequently, they tend to have reduced visual acuity (VA) along with severe photophobia and nystagmus, leading to vision impairment. Most of the population live in rural areas isolated from health care services with minimal knowledge of medical conditions or management options. People with albinism therefore remain poorly understood, often caught in a world of spiritual beliefs and superstition. Those who do seek assistance seldom receive optimum treatment or glasses because most rural 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 Dhanalakshmi Srinivasan Medical College, Perambalur. The Department of Ophthalmology enrolled 39 subjects satisfying the inclusion criteria. The diagnostic criteria for OCA patients included in the study, were those having ocular features of iris transillumination and retinal hypopigmentation in addition to depigmentation of the skin, hair and nails. Out of 39 participants, 5 were already using glasses previously. They were re-evaluated and prescribed new glasses with photochromatic lenses wherever required. A written informed consent was obtained from each participant or legal guardian involved in the study.

The study was approved by the Institutional Ethics Committee on Human Studies (IECHS) and the Institutional Research committee (IRC), adhering to the tenets of Declaration of Helsinki.

Demographic profile, history of chief complaints, previous use of low vision devices and use of any refractive correction were recorded. Unaided and aided visual acuity 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 pharmacological mydriasis.

Cycloplegic refraction and retinoscopy was performed 30 minutes after a three time instillation of a single drop of 1% Cyclopentolate eye drops, spaced 5 min apart. Participants who had emmetropia were excluded from the study.

Participants were interviewed over phone 3 weeks after glasses were dispensed. Compliance of the participants and tolerance towards usage of glasses was recorded as excellent (>75% of awake hours), good (50–75%), fair (26–50%), or poor (<25%) according to the information given by participants or legal guardians. They were asked unstructured open ended questions about the impact of glasses on their quality of life and activities of daily living.

Results

39 patients with OCA were included, mean age 20 years with 21 male and 18. The largest proportions of participants (40%) were between16 and 25yrs (Table 1).

Using world health organization classification based on best corrected distance visual acuity, 8%, 70%, 15% and 7% of 78 eyes had normal vision, moderate VI, severe VI and blindness respectively (Table 2).

Table 1: Distribution of participants by age and sex

Age (years)	Sex		Total	Percentage (%)
	Male (21)	Female (18)		
0-5	3	-	3	7.6
6-15	5	3	8	20.5
16-25	8	10	18	46.1
26-35	4	3	7	17.9
36-50	1	2	3	7.6
Total	21 (53.8%)	18 (46.1%)	39	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	4 (5.1)	6 (7.6)	Normal
<6/18-6/60	30 (38.4)	60 (76.9)	Moderate
<6/60-3/60	25 (35.8)	20 (25.6)	visual impairment
<3/60-PL	28 (35.8)	7 (8.9)	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; 78 eyes

Near visual acuity (meters)	N (%)	
	Presenting visual acuity	Corrected visual acuity
1	15 (19.2)	20 (25.6)
1.25	20 (25.6)	25 (32)
1.5	1(1.3)	1(1.3)
1.6	0	1(1.3)
2	24(30.8)	20(25.6)
2.25	0	3(3.8)
2.5	2(2.6)	0
3.2	1(1.3)	2(2.6)
4	4(5.1)	810.2
4.5	0	1(1.3)
5	2(2.6)	0
Total Mean VA	2.13±1.19	1.83±0.91

Table 4: Refractive error analysis

Refractive error	N (%)
Myopia	15 (19.2)
Hypermetropia	5 (6.4)
Myopic astigmatism	25 (32)
Hypermetropic astigmatism	33 (42.3)
Total	78 (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

Individuals with oculocutaneous albinism have visual impairment that compromises their social interactions as compared 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 S. Khanal *et al*. [13], myopic astigmatism was most common.

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 profile, consistent with the participants' age distribution, probably

reflects the reported normal reading ability [19, 20] and intellectual development among persons with OCA [21]. However, this challenges the findings by Okoro *et al*. [22]. 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% 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 of pterygium, pingueculum and ptosis were also 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, we highly recommend timely, unrestricted access to eye care services, provision of appropriate visual aids and management of co-existent ocular conditions, along with the need for creation of awareness amongst albinos regarding the same. 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.

References

1. Summers CG, Oetting WS, King RA. Diagnosis of oculocutaneous albinism with molecular. Am J Ophthalmol. 1996;121(6):724-6.
2. Kamaraj B, Purohit R. Mutational analysis of oculocutaneous albinism: a compact review. Biomed Res Int. 2014;2014:905472. [PMID: 25093188].
3. Passmore LA, Kaesmann-Kellner B, Weber BH. Novel and recurrent mutations in the tyrosinase gene and the P gene in the German albino population. Hum Genet. 1999;105:20010. [PMID: 10987646].
4. Gargiulo A, Testa F, Rossi S, Di Iorio V, Fecarotta S, De Berardinis T, *et al*. Molecular and clinical characterization of albinism in a large cohort of Italian patients. Invest Ophthalmol Vis Sci. 2011;52:1281-9.
5. Wei AH, Yang XM, Lian S, Li W. Genetic analyses of Chinese patients with digenic oculocutaneous albinism. Chin Med J (Engl). 2013;126:226-30. [PMID: 23324268].
6. Wilk MA, McAllister JT, Cooper RF, Dubis AM, Patitucci TN, Summerfelt P, *et al*. Relationship between foveal cone specialization and pit morphology in albinism. Invest Ophthalmol Vis Sci. 2014;55:4186-98.
7. Abadi R, Pascal E. The recognition and management of albinism. Ophthalmic Physiol Opt. 1989;9:3-15.
8. Carden SM, Boissy RE, Schoettker PJ, Good WV. Albinism: modern molecular diagnosis. Br J Ophthalmol. 1998;82:189-195.
9. Gronskov K, EK J, Brondum-Nielsen K. Oculocutaneous albinism. Orphanet J Rare Dis. 2007;2:43.
10. Biswas S, Lloyd IC. Oculocutaneous albinism. Arch Dis Child. 1999;80:565-569.
11. Spedick MJ, Beauchamp GR. Retinal vascular and optic nerve abnormalities in albinism. J Pediatr Ophthalmol Strabismus. 1986;23:58-63.
12. Bibbi Abruzzini. Feature: Nepal's albinos caught between reality, myth', Follow Xinhuanet. (http://news.xinhuanet.com/english/world/2014-10/09/c_133702376.htm)
13. Safal Khanal, Amrit Pokhrel, Himal Kandel. Visual deficits in Nepalese patients with oculocutaneous albinism. Journal of Optometry. 2016;9:103-9.
14. Eballe AO, Mvogo CE, Noche C, Zoua MEA, Dohvoma AV. Refractive error in Cameroonians diagnosed with complete oculocutaneous albinism. Cli Ophthalmol. 2013;7:1491.
15. Kromberg JG, Castle D, Zwane EM, Jenkins T. Albinism and skin cancer in Southern Africa. Clinical Genetics. 1989;36:43-52.
16. King RA, Creel D, Cervenka J, *et al*. Albinism in Nigeria with delineation of new recessive oculocutaneous type. Clinical Genetics. 1980;17:259-270.
17. Hong ES, Zeeb H, Repacholl MH. Albinism in Africa as a public health issue. BMC Public Health. 2006;6:212.
18. Cullinan TR. The epidemiology of visual disability. Studies of visually disabled people in the community. HSRU Report. 1977;28:136-137.
19. Collins B, Silver J. Recent experiences in management of visual impairment in Albinism. Ophthalmic Genetics. 1990;11(3):225-228.
20. National Organization of Albinism and Hypopigmentation (NOAH). Information for parent and children with Albinism. http://www.albinism.org Accessed. 2010 Dec 28.
21. Mohamed AF, El-Sayed NS, Seifeldin NS. Clinico-epidemiologic features of oculocutaneous albinism in northeast section of Cairo—Egypt. Egyptian Journal of Medical Human Genetics. 2010;11:167-172.
22. Okoro AN. Albinism in Nigeria. British Journal of Dermatology. 1975;92(5):485-492.
23. Jill Anderson *et al*. Efficacy of spectacles in persons with albinism. American Association for Pediatric Ophthalmology and Strabismus. 2004;8:515-20.