



ORIGINAL ARTICLE

Patterns of refractive errors in albino patients in pediatric age group.

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ABSTRACT... Objective: To evaluate the patterns of refractive errors in albino patients in pediatric age group and to find out the most common refractive error in these patients. **Study Design:** Cross-sectional study. **Setting:** Eye OPD of The Children's Hospitals and The Institute of Child Healths, Lahore and Multan. **Period:** July 2021 to December 2021. **Material & Methods:** Between the ages of 02 to 12 years. The participants presented with oculocutaneous albinism (OCA) were included and the participants with ocular pathologies like congenital cataract, retinopathy of prematurity, buphthalmos etc were excluded. Cycloplegic refraction was done to evaluate the refractive errors and funduscopy was done to find out ocular pathology. Non-probability convenient technique was used. Chi square test was used for statistical analysis. **Results:** This study recruited 22 patients with OCA between the ages of 02-12 years (mean age: 6.86 ± 3.385), 13 were male patients and 09 were female patients. 19 patients (86.36 %) had visual acuity (VA) fix and follow the light and 03 (13.64 %) patients were not able to fix and follow the light. Most of the patients have moderate type of refractive error. 06 patients (27.3%) had mild refractive error, 09 patients (40.9%) had moderate and 07 patients (31.8%) had severe refractive error. 13 patients (59.1%) showed astigmatism, 08 patients (36.4%) showed hypermetropia and only 01 patient (4.5%) showed myopia. Nystagmus was present in all 22 patients. **Conclusion:** This study suggested that astigmatism was the most common refractive error in OCA patients followed by hypermetropia. If refractive errors are not managed timely it will affect their daily life activities and increase the burden on the community.

Key words: Nystagmus, Oculocutaneous Albinism (OCA), Photophobia, Refractive Errors, Strabismus.

INTRODUCTION

Albinism is a congenital, genetic heterogeneous group of disorders characterized by the defect in synthesis of melanin pigment that gives color to the eyes, hairs and skin. Melanin synthesis is a highly controlled and proficient biochemical process that takes place in specific ectodermal derived cells, known as melanocytes.¹ OCA is the form of albinism in which melanin pigment is present in reduced amount or absent in hair, skin and eyes. Clinical presentation include vision defect and high vulnerability to skin cancer. Genetically, albinism can manifest via many inheritance patterns, although autosomal recessive albinism is the most common mode.²

The main cause of OCA is mutation in TYR, TYRP1 and SLC45A2 gene. OCA occur in non-syndromic

and syndromic classification.³ People with OCA1 are presented with white hair and very pale skin at birth but with the passage of time after 1-3 years skin and hair may grow some pigment. Iris color is blue which may be changed to green or brown. People with OCA2 have colored hair and skin color is creamy white or tan. OCA3 is present in African individuals who have reddish brown skin and reddish hair. Iris become hazel to light brown. In OCA4, skin is creamy colored or tan to normal. Hair and skin color are correlated to each other. Iris color is blue, brown or hazel. Hair may silvery white to yellow to blonde. People with OCA5 have white skin color and golden hair. Clinical presentation of OCA6 in humans is highly variable with milder to hypopigmented skin, light/ blonde brown hair, and reduced to no pigmentation in the iris^{4,5} Other ocular characteristics include

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reduced vision, nystagmus, iris trans-illumination, and foveal hypoplasia. In OCA7, hair are blonde to dark brown but skin color is light.⁶

There are many syndromes of albinism which are linked with systemic associations like Chediak-Higashi syndrome in which patients are presented with hematological irregularity, lack of skin, hair and eyes color, lymphoproliferation and periodic infections. It's very severe disease appears in childhood. Hermansky Pudlak syndrome is a tyrosinase positive type of OCA. It is a lysosomal storage disease with platelets abnormalities, lung fibrosis and in some cases kidney disease occur. In Waardenburg syndrome ocular features are hypochromic irides with total or segmental heterochromia and choroidal discoloration while its systemic features are poliosis, deafness and sometime limb and neurological anomalies also occur.⁷

Albinism can affect individuals of all ethnic group, approximately 1 in 17000 people have one kind of albinism. This prevalence is minimum amongst Asians.⁸ Reduced VA (20/60 to 20/400), refractive errors, congenital nystagmus, hypo-pigmented fundi, hypo-pigmented iris, foveal hypoplasia, reduced pigmentation of retinal epithelial layer, color vision impairment, prominent photophobia, strabismus, reduced stereoscopic vision, misrouting of optic nerves comprises of an excessive crossing of fibers in optic chiasma are the clinical manifestations of OCA.⁹ The delayed diagnosis can lead to high rate of complications such as due to decrease vision reduced to work opportunities, due to uncorrected refractive errors reading difficulties occur.¹⁰

Refractive errors (ametropia) are wider in albinism because of impaired emmetropization process due to deficiency of ocular structures hence the main feature of OCA is hypo pigmented fundi, hypo pigmented irides which leads to sensitivity to bright light and iris illumination.¹¹ These patients commonly have refractive errors such as myopia, hypermetropia and astigmatism. Ametropia is a condition in which equal light beams coming from limitlessness neglects to concentrate on the retina i.e., the secondary principal focus of the eye does

not fall on retina.¹² Myopia or short sightedness is the type of refractive error in which equal light rays that are coming from infinity concentrate in front of the perceiving layers of retina when the accommodation is at rest. Hypermetropia or long sightedness is the type of refractive errors in which equal light rays that are coming from infinity concentrate behind the perceiving layers of retina when the accommodation is at rest. Astigmatism is the type of refractive error in which a point focus of light cannot produce a punctate image on the retina and there is unequal refraction of light in different meridians.

Bifocal glasses may give adequate help for refractive errors. Dark, tinted or photochromic glasses may protect the eyes from photophobia and discomfort of the eyes. Nystagmus can be better with contact lenses and extraocular muscle surgery. Head posture helps to decrease the nystagmus. In cases of amblyopia, patching the child's good eye and forcing to use the lazy eye helps to improve VA. Sun screens are suggested with minimum sun protection factor of 15 for skin. No dietary or exercises will stop or alter the clinical demonstration of albinism.¹

Poor vision in OCA patients secondary to refractive errors affects performance in school and in daily life resulting in a negative impact on the present and future career with increased burden on the community. The planning of eye care programs requires data on the prevalence and distribution of refractive errors. Limited data on distribution of refractive errors in OCA patients is available required to plan strategies for the increasing awareness and education of the OCA patients and their parents about refractive error correction with spectacles and to transform this increased awareness to actual utilization of services. This study aimed to report the burden of VA loss due to refractive errors in OCA patients.

MATERIAL & METHODS

This cross-sectional study recruited all patients who visited eye OPD of The Children's Hospitals and The Institutes of Child Health, Lahore and Multan from July 2021 to December 2021, between the ages of 02 to 12 years. The article

was approved from institutional ethical committee vide Ref. No. 2021-303-CHICH, dated 16-06-2021. The participants presenting with OCA were included and the participants with ocular pathologies like congenital cataract, retinopathy of prematurity, buphthalmos etc were excluded. After taking consent from the child's attendant, cycloplegic refraction was carried out to evaluate the refractive errors and funduscopy was done to find out ocular pathology. Non-probability convenient technique was used and Chi square test was used for statistical analysis. A p-value of greater than 0.05 was considered insignificant whereas a p-value less than 0.05 was considered significant.

RESULTS

A total of 22 patients were recruited for this study between the ages of 02 to 12 years (mean age was 6.86 ± 3.385 years), 13 (59.1 %) were male and 9 (40.9 %) were female as shown in Table-I. Nystagmus was present in all 22 patients.

Gender	Frequency (%)
Male	13 (59.1%)
Female	09 (40.9%)
Total	22 (100%)

Table-I. Gender of participants

Table-II. shows visual assessment of all patients. 19 patients (86.36 %) were able to fix and follow the light and 3 patients (13.64 %) were unable to fix and follow the light out of 22 patients.

Visual Assessment	Frequency (%)
Able to Fix and Follow light	19 (86.36%)
Unable to Fix and Follow light	03 (13.64%)
Total	22 (100%)

Table-II. Visual assessment of participants

Table-III shows the severity of refractive errors. The refractive error was categorized as mild

($\leq 1D$), moderate ($> 1D$ to $4D$) and severe ($> 4D$). Most of the patients have moderate type of refractive error.

Severity of Refractive Error	Frequency (%)
Mild	06 (27.3%)
Moderate	09 (40.9%)
Severe	07 (31.8%)
Total	22 (100%)

Table-III. Severity of refractive error

Table-IV shows types of refractive error. Out of 22 patients, 13 (59.1%) patients showed astigmatism, 08 (36.4%) patients showed hypermetropia and 01 (4.5%) patient showed myopia.

Type of Refractive Error	Frequency (%)
Astigmatism	13 (59.1%)
Hypermetropia	08 (36.4%)
Myopia	01 (04.5%)
Total	22 (100%)

Table-IV. Type of refractive error

Table-V shows the cross tabulation between type of refractive errors and severity of refractive errors. 13 patients had astigmatism, 08 patients had hypermetropia and 01 patient had myopia.

In Table-VI chi square test was used and p-value indicated that there was no significance between type of refractive errors and gender, both male as well female had refractive errors.

DISCUSSION

Patients with OCA always face emotional and social problems due to impairment of vision. This condition leads to stigmatization.⁶ Patients with OCA have decreased vision, high refractive errors and photophobia.⁸ Refractive errors have high association with OCA.¹³

Type of Refractive Error	Refractive Error Severity			Total	Asymptomatic Significance (2-sided)
	Mild	Moderate	Severe		
Astigmatism	5	6	2	13	0.060
Hypermetropia	0	3	5	8	
Myopia	1	0	0	1	
Total	6	9	7	22	

Table-V. Cross tabulation between type of refractive errors and severity of refractive errors

Gender	Type of Refractive Error	Refractive Error			Total	Asymptomatic Significance (2-sided)
		Mild	Moderate	Severe		
Females	Astigmatism	3	0	1	4	0.186
	Hypermetropia	0	1	3	4	
	Myopia	1	0	0	1	
	Total	4	1	4	9	
Males	Astigmatism	2	6	1	9	0.243
	Hypermetropia	0	2	2	4	
	Myopia	0	0	0	0	
	Total	2	8	3	13	

Table-VI. Cross tabulation between type of refractive error and gender

In our study we concluded that most common refractive error in patients with OCA was astigmatism followed by hypermetropia showing almost same results as mentioned in previous studies.

Altinbay in 2020 reported 31 OCA patients with age range 05-58 years presented with decreased VA. VA improvement in these patients was mild to moderate in myopia but noticed a little improvement in hypermetropia above +1.50D. Astigmatism was with the rule and most commonly detected refractive error was hypermetropia.¹⁴ In our study we also concluded that most common refractive errors were astigmatism and hypermetropia. So there was no significant difference between our study and this study.

Sayed, Abdellah and Kamel in 2021 published a study in which mean age was 5.16 ± 3.4 years in albino group of 82 patients. Their study showed hypermetropia and astigmatism were most common refractive errors. This study showed relatively accurate results due to large population size. They used auto refractometer for cycloplegic refraction and determination of axes of astigmatism. In comparison to this study, our study showed almost similar results except we use retinoscope rather than auto refractometer for cycloplegic refraction and determination of accurate axes of astigmatism.¹⁵

Regarding refractive errors, some studies revealed that hypermetropia was most commonly present, other studies showed myopia was most frequent while few others reported equal prevalence of both refractive errors in OCA patients.¹⁴ Eballé AO in 2013 conducted a study in Cameroonians

population. 75 eyes in 35 patients were examined. Mean age in this study population was 12.30 ± 7.76 years. 28.6% were having myopia and 40% showed myopic astigmatism, 14.3% showed hypermetropia, 14.3% showed hypermetropic astigmatism and 02.8% showed mixed astigmatism. Only this study showed that myopia was most commonly present than hypermetropia in OCA patients. Our study showed astigmatism and hypermetropia as common refractive errors. This difference could be due to elder age group, secondly they used their accommodation hence myopia was most frequently present.¹⁶ Our study is limited by the small sample size.

CONCLUSION

This study indicated that astigmatism was the most common refractive error in OCA patients followed by hypermetropia. Refractive errors associated with OCA may lead to other ocular problems like strabismus and photophobia if not corrected properly. If refractive errors are not managed timely it will affect their daily life activities and increase the burden on the community. The need of the hour is to increase the awareness of refractive errors, wearing of glasses as well as improving the infrastructure in the hospitals for managing this problem.





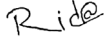
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REFERENCES

1. Yousaf S, Sethna S, Chaudhary MA, Shaikh RS, Riazuddin S, Ahmed ZM. **Molecular characterization of SLC24A5 variants and evaluation of Nitisinone treatment efficacy in a zebrafish model of OCA6.** Pigment Cell Melanoma Res. 2020; 33:556–565. Doi.org/10.1111/pcmr.12879.

2. Tomita Y, Suzuki T. **Genetics of pigmentary disorders, American Journal of Medical Genetics.** 2004; 131C (1): 75–81. Doi. org/10.1002/ajmg.c.30036.
3. Schidlowski L et al. **Non-syndromic oculocutaneous albinism: Novel Genetic Variants and Clinical follow up of a Brazilian Pediatric Cohort.** Front. Genet. 2020; 11:397. Doi. 10.3389/fgene.2020.00397.
4. Lasseaux E et al. **Molecular characterization of a series of 990 index patients with albinism.** Pigment Cell Melanoma Res. 2018 Jul; 31(4):466-474. doi: 10.1111/pcmr.12688.
5. Veniani E et al. **Detection of the first OCA6 Italian patient in a large cohort of albino subjects.** J Dermatol Sci. 2016 Mar; 81(3):208-9. Doi: 10.1016/j.jdermsci.2015.11.012.
6. Kromberg JGR, Manga P, Kerr R. **Children with oculocutaneous albinism in Africa: Characteristics, challenges and medical care.** S Afr J Child Health. 2020; 14(1):50-4. Doi.org/10.7196/SAJCH.2020.v14.i1.1608.
7. Salmon JF, Kanski JJ. Kanski's. **Clinical Ophthalmology a systematic approach.** In Hereditary Fundus Dystrophies. 9th ed. Edinburgh: Elsevier, 2020. 614-52.
8. Omar R, Idris SS, Meng CK, Knight VF. **Management of visual disturbances in albinism: A case report.** J Med Case Reports. 2012, 6:316, 01-4. Doi.org/10.1186/1752-1947-6-316.
9. Grønskov K, Ek J, Brøndum-Nielsen K. **Oculocutaneous albinism.** Orphanet J Rare Dis, 2007; 2:43. Doi. org/10.1186/1750-1172-2-43.
10. Federico JR, Krishnamurthy K. Albinism. In: **StatPearls. Treasure Island (FL): StatPearls Publishing; 2022.** PMID:30085560. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK519018/>.
11. Karlén E, Milestad L, Pansell T. **Accommodation and near visual function in children with albinism.** Acta Ophthalmol. 2019; 97(6):608-15. Doi: 10.1111/aos.14040.
12. Abrams D. **Duke-Elder's practice of refraction.** 10th ed. Edinburgh: Elsevier, 2019.
13. Wildsoet CF, Oswald PJ, Clark S. **Albinism: Its implications for refractive development.** Invest Ophthalmol Vis Sci. 2000; 41(1):1-7. PMID: 10634592.
14. Altinbay D. **Refractive errors and low vision rehabilitation in patients with oculocutaneous Albinism,** Pak J Ophthalmol. 2020; 36(2):150-155. Doi: 10.36351/pjo.v36i2.978.
15. Sayed KM, Abdellah MM, Kamel AG. **Analysis of the refractive profile of children with oculocutaneous albinism versus an age-matched Non-Albino Group.** Clin Ophthalmol 2021; 15: 73-78. Doi: [10.2147/OPTH.S286126](https://doi.org/10.2147/OPTH.S286126).
16. Eballé AO, Mvogo CE, Noche C, Zoua MEA, Dohvoma AV. **Refractive errors in Cameroonians diagnosed with complete oculocutaneous albinism.** Clin Ophthalmol. 2013; 7:1491-95. Doi.org/10.2147/OPTH.S38194.

AUTHORSHIP AND CONTRIBUTION DECLARATION

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2	Muhammad Ajmal Ch	Data collection, paper writing adn literature searching.	
3	Muhammad Ajmal Chaudhary	Study planning designing, manuscript writing and editing.	
4	Salah ud Din Arbi	Proof reading and editing.	
5	Rida Khalid	Data collection, paper writing and literature searching.	
6	Abida Hanif	Data entry and data analysis.	