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Impact of visual correction in people suffering with oculo-cutaneous Albinism in central region of Uttar Pradesh

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Abstract
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. Oculo-cutaneous 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.
Materials and Methods: This was a prospective community-based Study conducted at tertiary care Hospital in central region of Uttar Pradesh during 1 August 2016 to June 2020. The Department of Ophthalmology collected 100 People accepting enrolment voluntarily suffering with Albinism. The diagnostic criteria for OCA were: presence any two criteria as 1-Presence of Iris Transillumination 3-Depigmentation of the Skin 3-Retinal hypopigmentation 4- Depigmentation of the Hairs and Nails.
Results: 100 Patients with OCA were included, mean age 20 years with 60 (60%) males and 40 females (40%). The largest proportions of Participants (40%) were between16 to 25yrs. based on World Health Organization Classification for Visual impairment (VI) all Patients were categorized into Normal-Mild, Moderate Visual Impairment, Severe Visual Impairment, 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: oculo-cutaneous, albinism, hypo pigmentation, refractive error, vision

1. 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 Oculo-cutaneous Albinism [2, 4].
Oculo-cutaneous 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 Oculo-Cutaneous 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 Retino-geniculate 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 North Indian Professionals presume, they have poor potential for Visual improvement [12].

2. Materials and Methods
This was a Prospective Community based Study conducted at tertiary care Hospital in central region of Uttar Pradesh during 1 August 2016 to June 2020. The Department of Ophthalmology collected 100 People (60 male& 40 female) accepting enrollment voluntarily (200 Eyes). The diagnostic criteria for OCA were: presence of any two criteria 1-Presence of iris transillumination2-Retinal hypopigmentation and 3-Depigmentation of the Skin 4-Depigmentation Hair, and Nails. Out of 100 participants, 5 already had glasses. They were re-evaluated and prescribed new glasses with Photo chromatic 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 logMAR illiterate and literate charts (whichever appropriate) along with wet Retinoscopy and subjective Refraction. Extra ocular motility, Strabismus, fusion and Nystagmus were assessed. Anterior and posterior segment (Fundus) evaluation was performed under mydriasis.

2.1. Inclusion criteria
1-Cooperative & mentally sound
2-At least presence of two sign:
A--Presence of iris transillumination
B-Retinal hypo pigmentation
C-Depigmentation of the skin
D-Depigmentation of the hair and nails.

2.2. Exclusion criteria
1-Not cooperative, mentally retarded
2-Raised IOP
3-History of pigmentary Glaucoma
4-History of Trauma or any intraocular surgery.
5-Patient on topical Medication.
6-History of Inflammatory Eye Disease
7-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 hours), 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.

3. Results
100 Patients (200 Eyes) with OCA were included, mean age 20 years with 60 (60%) male and 40 female (40%). The largest proportions of participants (40%) were between16 to 25yrs Table 1 & Figure-1

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

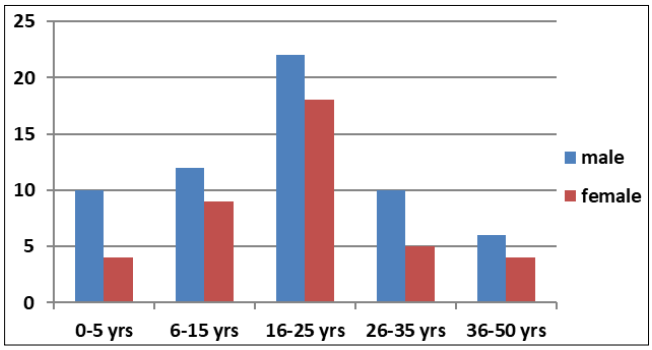

Fig 1: Distribution of participants by Age and Sex

Based on WHO criteria all patient considering distant vision were divided into four category-Normal to Mild Visual Impairment (5 %,) Moderate Visual Impairment (40%), Severe Visual Impairment (30%) and Blindness (25%) but when these Patient were improved with correct glasses the situation was improved as Normal to Mild Visual Impairment (7%) Moderate Visual Impairment (70%), Severe Visual Impairment (15%) and Blindness (8%). Table 2 & Figure-2

Table 2: Distance visual acuity of eyes at presentation and after correction

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

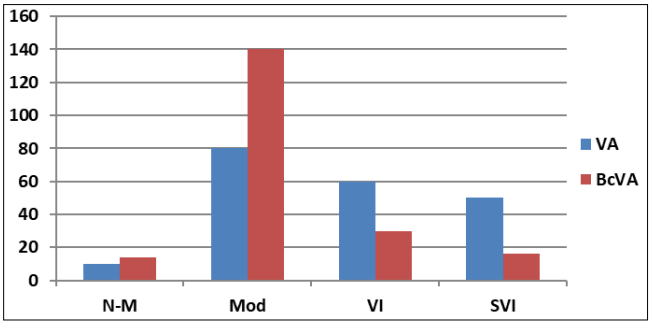

Fig 2: Distance visual acuity of eyes at presentation and after correction

As shown in Table 3 & Figure-3 Near Vision is also improved after prescription of correct glasses.

Table 3: Near visual acuity of eye by distance (meters) at presentation and after correction

Near visual acuity (meters)	Number (%)	
	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

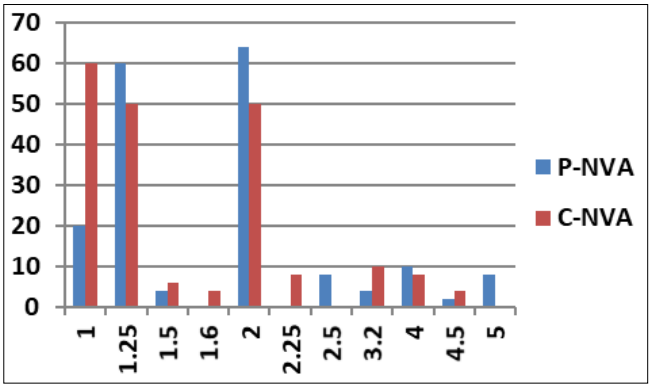

Fig 3: Near visual acuity of eye by distance (meters) at presentation and after correction

As shown in Table -4 and figure-4 Refractive error analysis the most of patients had Hypermetropic astigmatism (40%) but Hypermetropics were least in number (10%).

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)

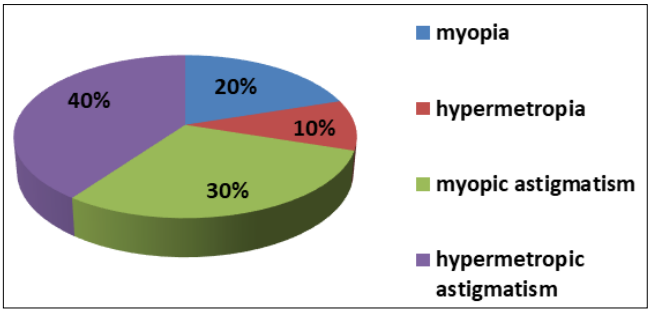

Fig 4: Refractive error analysis

Presence of strabismus was also a important manifestation of Oculo-cutaneous Albinism. It might be either for near or distance or for both for near and distance but both type of deviation improved after glass correction.

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

4. Discussion

Individuals with Oculo-cutaneous Albinism have Visual Impairment that compromises their Social interactions 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 [13] *et al*, 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 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% (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 VisualI 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.

5. 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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