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Ophthalmic Findings and Their Effect on Visual Function in Persons with Albinism in Southern Nigeria

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Authors' contributions

This work was carried out in collaboration between all authors. Author II designed the study and performed the statistical analysis, wrote the protocol and first draft of the manuscript. Authors CNP, AAO and BF managed the analyses of the study. Authors CNP and AAO managed the literature searches. All authors read and approved the final manuscript.

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ABSTRACT

Aim: To determine the pattern of ocular disorders in persons with albinism and how they affect visual function.
Methods: This was a descriptive cross-sectional study conducted over a five-month period on persons living with albinism in Southern Nigeria. The study participants were randomly selected during the monthly meetings of a support group known as The Albinism Foundation (TAF).Visual acuity and contrast sensitivity were assessed using the ETDRS visual acuity chart and Pelli Robson contrast sensitivity test chart. A comprehensive eye examination including dilated fundoscopy was also carried out to determine other ocular disorders. Data was analyzed using SPSS version 22 and statistical significance was set at a p-value ≤ 0.05.
Results: A total of 116 PWA (232 eyes) were examined. There were 44 (37.9%) males and 72 (62.1%) females. The age of the study subjects ranged from 5 to 56 years. Most eyes were visually impaired for both distance (n=228; 98.3%) and near vision (n= 224; 96.6%). Contrast sensitivity in most eyes (n=138; 59.5%) was subnormal. With refraction and Low Vision Aid (LVA), there was

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significant improvement of the mean VA by 2-3 acuity lines and 6 acuity lines respectively (p=0.000). All the examined eyes had fundus hypo-pigmentation, 91.4% (n=212) had iris trans-illumination, 86.2% (n=200) had nystagmus, and 34.4% (n=80) had strabismus. Nystagmus, strabismus and iris trans-illumination significantly (p=0.00) reduced visual acuity and contrast sensitivity.

Conclusion: Most study participants had reduced distance visual acuity and contrast sensitivity but with correction there was a significant improvement in vision. The presence of nystagmus, strabismus and iris trans-illumination were observed to contribute to the poor vision experienced by most persons living with albinism. Hence, early optical intervention and counselling is important in improving the quality of living of persons with albinism.

Keywords: Albinism; Nigeria; ocular albinism; ophthalmic findings; visual effect.

1. INTRODUCTION

Albinism is a hereditary condition characterised by a decrease or absence of pigment in the hair, eyes and skin of affected individuals resulting from disruption in melanin pigment synthesis, as a result of mutations in various enzymes and membrane proteins [1]. Albinism is broadly classified into two categories – Oculo-cutaneous albinism (OCA) and Ocular albinism (OA). In Oculo-cutaneous albinism, the hair, skin and eyes are affected while in Ocular albinism the features may be confined to the eyes and visual system [2]. Every variant of albinism affects every race and ethnicity with varying degrees of prevalence. In Northern Ireland, the prevalence of albinism is 1:4500 to 1:6000 [3] while in The United States of America, it is 1:18000 [4]. On the other hand, in Sub-Saharan Africa, the prevalence varies from 1:1000 in Zimbabwe [5] to 1:1400 in Tanzania, [6] and 1:3900 in South Africa [7]. In Nigeria, the prevalence of albinism is much lower at 1:15000 [8]; however, in West Africa, Nigeria has the largest population of persons with albinism [8,9] with most found in the southern part of the country [8].

Disruption in melanin synthesis has been proven to result in severe abnormality in the development of the visual system leading to several visual deficits including delayed visual maturation, nystagmus, iris trans-illumination, strabismus, impaired colour vision, visual impairment, reduced contrast sensitivity and stereoacuity [10,11]. Other ocular features associated with albinism include optic nerve hypoplasia, abnormal fovea and abnormal decussation of the optic chiasm [12].

In many countries including Nigeria, it has been established that most patients with albinism have abnormal distance and near visual acuity [13,14,15] and this is said to have a serious impact on the learning ability of children and the activities of daily living of adults [16]. Therefore current information about prevalence of visual impairment is essential in resource allocation and planning of prevention strategies in line with World Health Organisation's (WHO) global target to reduce the prevalence of avoidable visual impairment by 25% by 2019 from the baseline of 2010 [17].

The perception of contrast is very essential to vision, [18] yet only few studies have been done in Africa. In Nigeria [8,15] none of the studies on albinism evaluated impairment of contrast sensitivity which is an important disability in PWA. Different studies [19,20] have shown that difficulties in reading, orientation and mobility are more related to reduced contrast sensitivity than visual acuity. Assessment of contrast sensitivity provides valuable information in the early detection and monitoring of ocular diseases, as well as in evaluating the impact of therapy. Several studies [11,21] reported reduced contrast sensitivity function in most patients with albinism and stated that this contributed to visual impairment. Therefore, evaluation of contrast sensitivity is necessary to better understand the severity of visual impairment and aid in optimal prescription of magnifiers or spectacle correction.

2. METHODS

The study was a descriptive cross-sectional study carried out on 116 persons with albinism (PWA) recruited from The Albinism Foundation in Port Harcourt City, Rivers State, Nigeria. The study was approved by the Health Research Ethics Committee of The University of Port Harcourt Teaching Hospital and it followed the tenets of the Declaration of Helsinki. The study lasted five months and was conducted during their monthly meetings. All persons living with albinism who were five years and older with no ocular co-morbidities were included in the study.

A semi-structured pretested questionnaire specifically designed for the survey was used to record age, gender, relevant ocular and medical history, visual acuity, contrast sensitivity and clinical examination findings. Distance visual acuity was assessed using the Early Treatment Diabetic Retinopathy Study (ETDRS) chart placed at 4metres under standard illumination and recorded in Logarithm of the Minimum Angle of Resolution (logMAR) units while the near acuity was assessed using Radner Reading Chart at 40cm. Visual acuity was classified into 5 groups based on the WHO International Classification of Diseases 9 and 10. Pelli–Robson Chart at 1 m was used to measure contrast sensitivity (CS) [22] and recorded as log contrast sensitivity. A score of <1.65 log CS was considered to be visual impairment and < 1.05 log CS was considered to be visual disability [23]. A complete ophthalmic examination of the anterior segment and assessment of iris trans-illumination with dilated fundus examination for the macula was done. Foveal hypoplasia was then diagnosed based on the absence of foveal reflex/depression while macular transparency was diagnosed based on the prominence of choroidal vessels in the macula.

Data obtained was analyzed using the Statistical Package for Social Sciences (SPSS version 22, SPSS Inc., IBM, USA). Simple statistics such as means and frequencies were determined. Continuous variables were compared using independent sample t-test, while categorical parameters were analyzed with the Chi-square test. A p-value $\leq$ 0.05 was considered statistically significant.

3. RESULTS

Table 1 shows that most of the participants (N=78; 67.2%) were adults with a mean age of 34.23 ± 9.43 years while the number of participating children (those aged below 18 years) was 38 (32.8%) with a mean age of 9.47 ± 3.13 years. The age range was 5 – 56 years and there was a female preponderance in both categories with a male: female ratio of 1:1.63.

In Table 2, the frequency distribution of presenting visual acuity was analysed based on WHO classification of visual impairment. A total of 228 (98.3%) eyes had visual impairment for presenting distance vision while 224 (96.6%) eyes were visually impaired for near vision. Most of the participants had moderate visual impairment for presenting distance (n=172; 74.1%) and near acuity (n=162; 69.8%). Normal contrast sensitivity was noted in 94 (40.5%) eyes while 138 (59.5%) eyes had reduced contrast sensitivity. The number of eyes with visually impaired contrast sensitivity was 110 (47.4%) while visual disability based on contrast sensitivity was noted in 28 (12.1%) eyes.

Table 3 shows that the mean presenting distance and near VA significantly improved by 2-3 lines of the acuity chart with refraction (p=0.000) and by about 5-6 lines of the acuity chart with low vision aid (p=0.000). There was a strong correlation between presenting visual acuity and the corrected VA post refraction (R_S=0.785, p=0.00) and low vision aid (R_S =0.673, p=0.00).

As shown in Fig. 1, both eyes of all participants (232 eyes) had macular transparency (prominent choroidal vessels), fundus hypopigmentation and foveal hypoplasia. Nystagmus was present in 200 eyes (86.2%) while iris trans-illumination was seen in 212 eyes (91.4%). Strabismus was noted in only 80 (34.5%) eyes.

Table 4 shows that all the measures of visual function were affected by the presence of nystagmus, strabismus and iris trans-illumination. The mean logMAR distance visual acuity for participants with nystagmus (0.94±0.34) and iris trans-illumination (0.92±0.34) was found to be significantly worse (p = 0.000) by 2 or more lines when compared with the mean visual acuity of participants who didn't have nystagmus (0.52±0.20) or iris trans-illumination (0.42±0.17). In those with strabismus, it reduced significantly by about one line of the visual acuity chart (p=0.002). Similar trend was noted with near acuity measures. Contrast sensitivity was also significantly degraded by 2-3 triplets of letters in those with nystagmus (p=0.001) and strabismus (p=0.009) while in participants with iris trans-illumination; it was, significantly degraded by 4-5 triplets of letters (p= 0.000).

4. DISCUSSION

In many developing countries like Nigeria, people living with albinism have a myriad of visual challenges that affect their daily lives. In this study, the relationship between ocular findings and visual function was assessed. The mean presenting distance and near visual acuity was reduced with most participants presenting with moderate visual impairment for both distance and near vision. This is similar to the results by

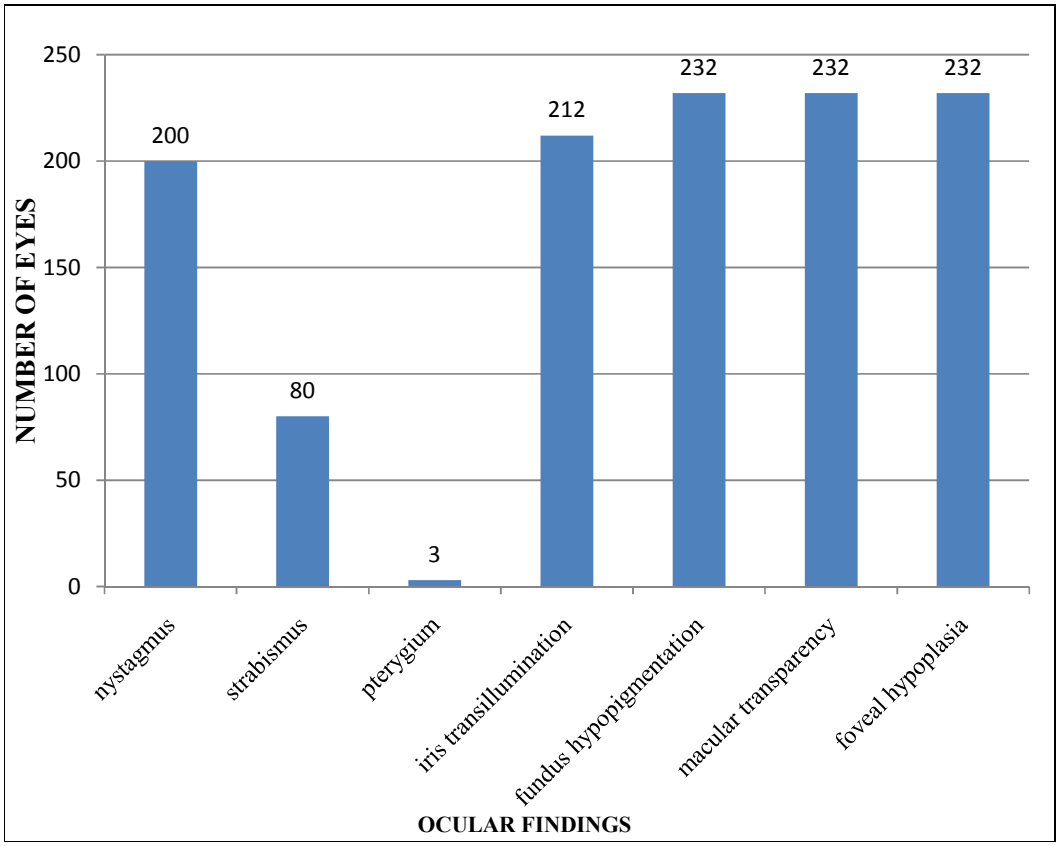

Fig. 1. Pattern of ocular findings in individual eyes

Udeh et al. [15] and Ajose et al., [24] in other parts of Nigeria. Other developing countries with similar socioeconomic status such as Nepal, [13] Malawi [25] and Cameroun [26] also had similar findings. It is said that the subnormal presenting visual acuity seen in persons with albinism may be due to delayed visual maturation, foveal hypoplasia, nystagmus, strabismus and iris trans-illumination. Anderson et al. in The USA [27] however reported a better mean uncorrected distance and near visual acuity in their study subjects. This is not surprising as there is easy availability of quality health care in The USA making it possible to access low vision aids (LVA). However, the same does not apply in Nigeria, where almost all medical expenses are paid by individuals, and with poverty and low socioeconomic status most PWAs can hardly afford any LVA. Amblyopia may also be a major contributor to the visual impairment observed as most of the participants had never used refractive correction and even amongst the few who had worn glasses, most only started wearing glasses for presbyopic correction. This could in part explain the better mean acuity reported in The USA study [27] where early eye check-up is encouraged and also readily available.

There was a noticeable rise in the number of eyes that had improved visual acuity after optical correction. Previous studies [13,15,26] both within Nigeria and elsewhere also reported significant improvement in presenting distance visual acuity after optical correction. In Nepal, Khanal et al. [13] recorded a mean improvement of 2 logMAR acuity lines after refraction; which is similar to this study; where there was an improvement of 2-3 logMAR acuity lines of mean distance and near vision. Despite the recorded improvement with refraction, there was still overall subnormal vision in participants and the reason may be because of the presence of varying degrees of foveal hypoplasia, in addition to possible development of amblyopia. However, with low vision aid, there was further improvement. This is similar to Udeh's study [15] where, even though the mean visual acuity improved after spectacle correction there was still marked impairment in vision. However with low vision aid an improvement in vision was

noted in their participants. These findings further buttress the need for promotion of early optical intervention especially low vision aids in persons with albinism as this would improve visual acuity, quality of life and therefore ability to achieve their full potential.

The participants, who were mostly adults, had diminished mean contrast sensitivity. This is not surprising as activities associated with contrast sensitivity such as driving at night or in the rain and navigation in unfamiliar surroundings (during business activities) are commonly undertaken by adults. Similar findings of impaired mean contrast sensitivity was noted in Chicago [21], Malawi [25] and Egypt [28]. Factors thought to be responsible for the degradation in contrast sensitivity include increased central retinal cone spacing, abnormal decussation of the visual pathway and glare from iris hypopigmentation, nystagmus and foveal hypoplasia.

Table 1. Distribution of children and adults

Age groups	Gender		Total No. (%)	Chi square (p value)
	Male no. (%)	Female no. (%)		
Below 18 years	16 (13.9)	22 (19)	38 (32.8)	0.209 (0.647)
18 years and older	28 (24)	50 (43.1)	78 (67.2)	
Total	44 (37.9)	72 (62.1)	116(100.0)	

Table 2. Distribution of presenting visual acuity and contrast sensitivity in 232 eyes

Visual Acuity Categories (LogMAR; Snellen quivalent)	Frequency	Percentage %
Distance visual acuity		
Normal vision (≥ 0.3; 6/12)	4	1.7
Mild VI (< 0.3 - ≥ 0.48; 6/12- 6/18)	8	3.4
Moderate VI (<0.48 - ≥1.0; 6/18- 6/60)	172	74.1
Severe VI (<1.0 - ≥ 1.3; 6/60- 3/60)	24	10.4
Blindness (<1.3 - ≥ 2.0; 3/60- NLP)	24	10.4
Total	232	100
Near visual acuity		
Normal vision (≥ 0.3; N6)	8	3.4
Mild VI (< 0.3 - ≥ 0.48; N6 –N10)	8	3.4
Moderate VI (<0.48 - ≥1.0; N10- N32)	162	69.8
Severe VI (<1.0 - ≥ 1.3; N32- N63)	30	13
Blindness (<1.3 - ≥ 2.0; N63- N320)	24	10.4
Total	232	100
Contrast sensitivity categories		
Normal CS (>1.65)	94	40.5
CS Visual Impairment (<1.65 - >1.05)	110	47.4
CS Visual Disability (<1.05)	28	12.1
Total	232	100

Legend: VI- Visual Impairment; CS- Contrast Sensitivity

Table 3. Comparison between presenting and corrected visual acuity in 228 eyes with visual impairment

Visual acuity		No. of eyes (%)	LogMAR(Snellen) mean ± SD	Mean difference	t test (P Value)	Correlation R_S (P value)
Distance visual acuity	Presenting	228	0.93± 0.32 (6/51)			
	Refraction	228	0.68± 0.23 (6/29)	0.25	13.35 (*0.00)	0.785(*0.00)
	LVA	188	0.35± 0.24 (6/13)	0.58	28.69 (*0.00)	0.673(*0.00)
Near visual acuity	Presenting	222	0.88 ±0.33 (N24)			
	Refraction	222	0.54 ±0.23 (N11)	0.34	18.42 (*0.00)	0.818(*0.00)
	LVA	136	0.32 ±0.12 (N6)	0.56	21.27 (*0.00)	0.611(*0.00)

*P value is based on paired samples T test and Pearson's correlation. *p value is statistically significant*

Table 4. Effect of anterior segment findings on mean visual function tests of 232 eyes

Anterior segment findings			Visual function tests		
		No. of eyes (%)	Mean± Std LogMAR distance VA	Mean± Std LogMAR Near VA	Mean ± Std contrast sensitivity
Nystagmus	Present	200 (86.2)	0.94 ± 0.34	0.91 ± 0.32	1.51 ± 0.57
	Absent	32 (13.8)	0.52 ± 0.20	0.49 ± 0.15	2.00 ± 0.30
	p value		*0.000	*0.000	*0.001
Strabismus	Present	80 (34.5)	1.02 ± 0.38	0.99 ± 0.32	1.38 ± 0.60
	Absent	152 (65.5)	0.80 ± 0.31	0.78 ± 0.33	1.69 ± 0.51
	p value		*0.002	*0.002	*0.009
Iris trans-illumination	Present	212 (91.4)	0.92 ± 0.34	0.89 ± 0.32	1.52 ± 0.55
	Absent	20 (8.6)	0.42 ± 0.17	0.43 ± 0.16	2.23 ± 0.47
	p value		*0.000	*0.000	*0.000

All study subjects had varying degrees of macular transparency, fundus hypopigmentation and foveal hypoplasia. Udeh et al. [15] reported similar posterior segment finding in her study. Other studies in Nepal [13] and The USA [10,29] also had similar results. The reason for the fundus hypopigmentation and foveal hypoplasia is because of reduced melanin pigmentation which is directly responsible for foveal hypoplasia and hypopigmentation. Nystagmus was present in most eyes, similar to studies in Nepal [13] and The USA [30]. However Udeh [15] in Enugu, Nigeria, reported 100% nystagmus in her study population. This higher percentage of nystagmus in Udeh's study may not be unconnected with the inclusion criteria for nystagmus which included minimal nystagmus detectable by slit lamp examination unlike this study where only those with obvious nystagmus were considered. Other differing studies [25,29] with similar methodology as Udeh's also reported universal presence of nystagmus. Underdevelopment of the fovea and delayed visual maturation seen in PWA is often considered to be responsible for the development of nystagmus. In almost all the eyes, varying degrees of iris trans-illumination was observed and this is due to the varying amount of melanin pigment present in every PWA. This is consistent with findings by Udeh at al. [15] but different from other studies [13,29] where iris trans-illumination was noted in all participants. The reason for the difference may be because the earlier studies [13,29] were all hospital-based.

In this study, strabismus was noted in 34.5% of eyes similar to other studies [31,32]. Strabismus is thought to result from abnormal decussation at the optic chiasm leading to misrouting of the visual pathway. A much lower frequency was however observed by Udeh et al. [15] and this may be because most participants in her study were more educated and therefore might have sought optical correction earlier thereby improving visual outcome. The mean distance and near visual acuity in eyes with nystagmus, iris trans-illumination or strabismus was significantly worse when compared with eyes that didn't have these features. This finding is similar to results by Khanal et al. [13] and Lee et al., [33] where nystagmus was found in all eyes with poor visual acuity. High refractive error from nystagmus and iris trans-illumination and abnormal binocular vision from strabismus is thought to play a role. This therefore implies that the absence of nystagmus, iris trans-illumination and to a minor degree, strabismus, is necessary to having normal visual acuity.

Contrast sensitivity was also significantly associated with iris trans-illumination, nystagmus and strabismus. It degraded by 2 to 3 triplets of letters in participants with nystagmus and strabismus and by 4 to 5 triplets in those with iris trans-illumination. Overall, Contrast sensitivity had the most association with iris trans-illumination; this further confirms the fact that glare effect from iris trans-illumination contributes largely to impaired contrast sensitivity.

5. LIMITATIONS OF THE STUDY

Proper equipments for fovea and macula assessment like OCT and flourescein angiography were unavailable due to financial constraint; therefore this study used dilated fundus examination for diagnosis.

6. CONCLUSION

This study shows that persons with albinism irrespective of age have varying degrees of visual impairment as a result of ocular disorders;

and this negatively affects their activities of daily living. The presence of nystagmus and iris trans-illumination is seen to play a role in the level of subnormal visual function. Therefore, continuous counselling to encourage early optical intervention from early childhood is necessary especially in the developing world. Increased provision of low vision centres and subsidized LVA will also aid in improving ocular health and general wellbeing of persons with albinism.

CONSENT

It is not applicable.

ETHICAL APPROVAL

The study was approved by the Health Research Ethics Committee of The University of Port Harcourt Teaching Hospital and it followed the tenets of the Declaration of Helsinki.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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