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ORIGINAL RESEARCH

Refractive errors in Cameroonians diagnosed with complete oculocutaneous albinism

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André Omgbwa Eballé[1,3]
Côme Ebana Mvogo[2]
Christelle Noche[4]
Marie Evodie Akono Zoua[2]
Andin Viola Dohvoma[2]

[1]Faculty of Medicine and Pharmaceutical Sciences, University of Douala, Douala, Cameroon, [2]Faculty of Medicine and Biomedical Sciences, University of Yaoundé I, Yaoundé, Cameroon; [3]Yaoundé Gynaeco-Obstetric and Paediatric Hospital, Yaoundé, Cameroon; [4]Faculty of Medicine, Université des Montagnes, Bangangté, Cameroon

Background: Albinism causes significant eye morbidity and amblyopia in children. The aim of this study was to determine the refractive state in patients with complete oculocutaneous albinism who were treated at the Gynaeco-Obstetric and Paediatric Hospital, Yaoundé, Cameroon and evaluate its effect on vision.

Methods: We carried out this retrospective study at the ophthalmology unit of our hospital. All oculocutaneous albino patients who were treated between March 1, 2003 and December 31, 2011 were included.

Results: Thirty-five patients (70 eyes) diagnosed with complete oculocutaneous albinism were enrolled. Myopic astigmatism was the most common refractive error (40%). Compared with myopic patients, those with myopic astigmatism and hypermetropic astigmatism were four and ten times less likely, respectively, to demonstrate significant improvement in distance visual acuity following optical correction.

Conclusion: Managing refractive errors is an important way to reduce eye morbidity-associated low vision in oculocutaneous albino patients.

Keywords: albinism, visual acuity, refraction, Cameroon

Introduction

Albinism is an inherited abnormality of melanin synthesis. Its clinical manifestations are related to the reduction or absence of pigmentation in the visual system and/or the skin and teguments.[1] Albinism is a universal condition that demonstrates an incidence of one case per 20,000 births.[2] Albinism is more common in black people, and its transmission is autosomal recessive in most cases.[3]

During the development of the optic system, melanin deficiency causes clinical manifestations such as foveal hypoplasia, strabismus, nystagmus, photophobia, and refractive errors.[2] This explains the low visual acuity that presents from birth. The cellular integrity of the fovea in albino patients is not well understood, but important anatomical differences most likely underlie the phenotypic variability of this disease and may also affect patient responsiveness to therapeutic intervention.[4]

McAllister et al[4] reported that foveal morphology and cone specialization are variable in albino patients (based on examinations of the foveal pit and determination of photoreceptor outer segment elongation). According to Mohammad et al,[5] the size of the outer segment of the photoreceptor is the strongest predictor of best corrected visual acuity in albino patients.

Correspondence: André Omgbwa Eballé
Ophthalmology Unit, Yaoundé Gynaeco-Obstetric and Paediatric Hospital,
PO Box 4362, Yaoundé, Cameroon
Tel +237 996 544 68
Fax +237 2222 4419
Email andyeballe@gmail.com

Refractive errors are common in albino patients, and relatively high values are reported in the literature.[6,7] In the African literature, very little data exists on the vision of oculocutaneous albino patients. Other authors[8,9] studied ophthalmic disorders in all types of albinos between 2001–2007 in Douala, Cameroon. Here, our study focuses only on refractive errors in patients diagnosed with complete oculocutaneous albinism. The aim of this study, which was carried out at the ophthalmology unit of Yaoundé Gynaeco-Obstetric and Paediatric Hospital, Yaoundé, Cameroon, was to determine the refractive states of albino patients with complete oculocutaneous albinism and any correlations with vision.

Methods

This retrospective study was carried out between March 1, 2003 and December 31, 2011. Data were collected from the medical records of all patients. All patients with complete oculocutaneous albinism $\geq$ 5 years of age were included. Patients $<$ 5 years of age were excluded because it is difficult to obtain accurate measurements of their visual acuities. Patients with ocular albinism and the yellow mutant forms of oculocutaneous albinism were also excluded.

Patients were not classified as tyrosinase-positive or tyrosinase-negative due to the fact that tyrosinase hair bulb incubation tests were not performed as part of this study. Clinical evaluations included the following: evaluation of symptoms and past medical history; visual acuity testing using Snellen's tumbling E chart; and external clinical examination and slit-lamp examination of the anterior and posterior segments of the eyes, respectively. Intraocular pressure was measured using a non-contact tonometer (Topcon Medical Systems, Paris, France).

Diagnosis was based on the presence of the following conditions: iris transillumination, foveal hypoplasia, and retinal hypopigmentation in association with depigmentation of the skin, hair, and nails.[10,11] Cycloplegic automatic refraction was performed using the RM-8000D system (Topcon Medical Systems). Cycloplegia was achieved with 0.5% tropicamide (Mydriaticum®) and 0.5% cyclopentolate (Skiacol®). A drop of each was instilled alternately at 5-minutes intervals. A total of 3 instillations for each drug were made. Automatic refraction was performed for 45–60 minutes after the last instillation.

In this study, myopia is defined as spherical ametropia in which parallel light rays coming from infinity focus on a plane in front of the retina; hypermetropia refers to spherical ametropia in which parallel light rays coming from infinity focus on a plane behind the retina.[12]

Astigmatism refers to an optical system that does not produce a point image from a point object, as opposed to a stigmatic system which does produce a point image from a point object. Direct astigmatism is known as "with-the-rule" astigmatism when the radius of curvature of the vertical meridian is less than that of the horizontal meridian. In indirect or "against-the-rule" astigmatism, the radius of the curvature of the horizontal meridian is less than that of the vertical meridian. In oblique astigmatism, the principal meridians are perpendicular to each other and situated at 45° and 135°. Astigmatism is considered "simple" when one focal line is on the retina, but this could be diagnosed as simple myopic or simple hypermetropic astigmatism depending if the other focal line is in front of or behind the retina, respectively. If both focal lines are found on the same side of the retina, this could be diagnosed as myopic or composed hypermetropic astigmatism. Mixed astigmatism is diagnosed when both focal lines are found on either side of the retina.[12]

The following variables were analyzed: age, sex, uncorrected distance visual acuity, automatic refraction, and best corrected distance visual acuity (CDVA). Ametropia was classified as myopia, hypermetropia, myopic astigmatism, hypermetropic astigmatism, or mixed astigmatism in order to avoid determination of the spherical equivalent.

There are four levels of visual function according to the International Classification of Diseases-10: normal vision (CDVA $\geq$ 3/10); moderate visual impairment (1/10 $\leq$ CDVA $<$ 3/10); severe visual impairment (1/20 $\leq$ CDVA $<$ 1/10); and blindness (CDVA $<$ 1/20). Moderate and severe visual impairment were considered "low vision." Low vision together with blindness represents all types of visual impairments.[13] This definition was used to classify visual impairment.

Table 1 Distribution of cases by age

Age (years)	n	Percentage (%)
5–15	25	71.43
16–25	8	22.86
26–37	2	5.10
Total	35	100.00
Summarized measurements		**Value**
Mean (years)		12.30
Standard deviation		7.76
95% CI for the mean		10.45–14.15
Minimum (years)		5
Maximum (years)		37

Abbreviations: n, number; CI, confidence interval.

Table 2 Distance visual acuity analysis

Distance VA	Uncorrected n (%)	Corrected n (%)	WHO definition
VA > 3/10	0 (0)	14 (20%)	No visual impairment
3/10 > VA ≥ 1/10	49 (70%)	52 (74.3%)	Moderate visual impairment
1/10 > VA ≥ 1/20	15 (21.42%)	3 (4.28%)	Severe visual impairment
VA <1/20	6 (8.58%)	1 (1.42%)	Blindness
Mean distance VA	0.11 ± 0.05	0.15 ± 0.08	Moderately low vision

Abbreviations: VA, visual acuity; n, number; WHO, World Health Organization.

Patients were divided into two groups: one group consisted of patients whose visual acuity improved following optical correction (group 1), and the other consisted of patients who did not demonstrate improvement (group 2).

Microsoft Excel 2007 (Microsoft Corporation, Redmond, WA, USA), CS Pro 3.3 (Informer Technologies, www.informer.com), and SPSS (IBM Corporation, Armonk, NY, USA) were used to perform all statistical analyses. Student's *t*-test was used to compare the means between the two groups, and analysis of variance was used to analyze multiple groups together. The Chi-square test, Pearson linear correlation test, and multivariate logistic regression were used to determine any relationships between variables. Tests were considered statistically significant when P-values < 0.05 were determined.

Results

During the study period, 70 eyes in 35 patients were examined. More males (n = 20; 57%) than females (n = 15; 47%) were included, demonstrating a male:female sex ratio of 1.33.1. The mean age of the study population was 12.30 ± 7.76 years. Twenty-five patients (71.43%) were between 5–15 years of age, and the oldest patient was 37 years old (Table 1). Uncorrected distance visual acuity was <1/20 in six eyes (8.58%), <3/10 but ≥1/10 in 49 eyes (70%), and <1/10 but ≥1/20 in 15 eyes (21.42%). CDVA of one eye (1.42%) was <1/20, thereby meeting the World Health Organization's definition for blindness.[13] Fifty-two eyes (74.3%) had CDVA < 3/10 but ≥1/10 and were classified as moderate visual impairment. However, three eyes (4.28%) had CDVA < 1/10 but ≥1/20 and were classified as severe visual impairment. Fourteen eyes (20%) had a CDVA ≥ 3/10 (Table 2).

According to the examinations that used the Snellen's tumbling E chart, the mean uncorrected distance visual acuity was 0.11 ± 0.05. Mean visual acuity rose to 0.15 ± 0.08 with optical correction. There was a positive linear correlation between uncorrected and corrected visual acuities, demonstrating a Pearson linear correlation coefficient of 0.727 (P = 0.000; Table 3). Myopic astigmatism was the most common refractive error (n = 28; 40%), followed by myopia (n = 20; 28.6%). Hyperopia and hyperopic astigmatism were diagnosed in ten eyes each (14.3%), and mixed astigmatism was diagnosed in two eyes (2.8%; Table 4). All cases of astigmatism were diagnosed as "with-the-rule" and ranged from 0.50–6.50 diopters, demonstrating a mean of 3.62 ± 1.8 diopters. Subjective refraction was analyzed in all patients, leading to improvement in distance visual acuity by one or two lines on the Snellen's tumbling E chart in 51.42% of patients (n = 18 of 35 patients; 36 eyes were analyzed). However, distance visual acuity could not be improved in 48.58% of patients (n = 17) diagnosed with amblyopia. The mean age of the patients whose distance visual acuity improved with optical correction was 13 ± 8.99 years (group 1 patients), in comparison with 11.56 ± 6.26 years for patients whose distance visual acuity could not be improved using optical correction (group 2 patients; P = 0.442; Table 5). The degree of refractive error did not influence corrected visual acuity – instead, the type of refractive error statistically influenced corrected visual acuity. Myopic and hyperopic patients demonstrated improvements in distance visual acuity following optical correction, while astigmatic patients did not demonstrated improvement regardless of the type of the astigmatism (Table 6).

Discussion

Visual impairment in patients with complete oculocutaneous albinism worsens the emotional and social problems associated with this condition. This leads to greater stigmatization and

Table 3 Analysis of uncorrected and corrected distance visual acuity measured in all eyes (n = 70)

	Minimum	Maximum	Median	Mean ± standard deviation	95% confidence interval	*P**
Uncorrected	0.02	0.20	0.10	0.11 ± 0.05	0.09–0.12	0.000
Corrected	0.02	0.30	0.10	0.15 ± 0.08	0.13–0.17	

Note: *Determined using Student's *t*-test.
Abbreviation: n, number.

Table 4 Refractive errors analysis

Refractive error	n	%	Mean power (D)	Standard deviation	95% confidence interval	*P**
Myopia	20	28.6	7.74	4.41	5.67–9.80	0.000
MA	28	40	3.72	1.47	3.15–4.29	
Hypermetropia	10	14.3	3.80	2.96	1.68–5.92	
HA	10	14.3	3.58	2.17	2.02–5.13	
Mixed astigmatism	2	2.8	2.75	0.35	–0.43 to 5.93	
Total	70	100	4.83	3.38	4.03–5.64	

Note: *Determined using ANOVA.
Abbreviations: n, number; D, diopters; MA, myopic astigmatism; HA, hypermetropic astigmatism.

poor integration into society because albinism is still considered taboo in Cameroon despite its relatively common worldwide distribution and incidence of one case per 20,000 births.[2]

Refractive errors are common in oculocutaneous albino patients. Some studies report hypermetropia as the most common refractive error,[2,7,14] while others report myopia.[6,15] In a recent study of the refractive profiles of patients with oculocutaneous albinism, Yahalom et al[16] reported astigmatism and hypermetropia as the most common refractive errors. In this study, myopic astigmatism was the most common, representing 40% of patients. This is similar to the results reported by Mvogo et al[8] who reported that 61.9% of albino patients develop myopic astigmatism. This is contrary to studies on the general Cameroonian population that report hypermetropia as the most common refractive error.[17–19]

Astigmatism in albino patients, as reported in the literature, is generally classified as with-the-rule and high-power,[7,20–22] and our results are in agreement with this. Patients diagnosed with with-the-rule astigmatism demonstrated a mean power of 3.62 diopters. Optical correction did not improve distance visual acuity, regardless of the type of astigmatism. This is contrary to myopia, in which there was a moderate but statistically significant improvement in distance visual acuity. Multivariate logistic regression modeling (Table 5) shows that oculocutaneous albino patients with myopic astigmatism are about four times less likely to demonstrate improvement in distance visual acuity following correction in comparison with myopic patients. Similarly, patients diagnosed with hyperopic astigmatism are nearly ten times less likely to have their distance visual acuity improved in comparison with myopic oculocutaneous albino patients.

These improvements in the distance visual acuities of several patients with eyeglasses corroborate the data reported in the literature.[10,23] Although the improved visual acuities of these patients largely remain below normal, it does, however, improve the quality of life of these patients. Albinism is an important cause of low vision, accounting for 5% of low-vision cases worldwide.[24] Low vision in oculocutaneous albino patients can also be due to nystagmus or low macular cone density.[25–27] Considering the fact that the majority (71.43%) of our patients were between 5–15 years of age, albinism can be considered one of the leading causes of childhood amblyopia and educational retardation in Cameroon. In a study carried out in 2000, Bella et al[28] reported that 7.5% of cases of blindness among children aged 0–5 years are associated with oculocutaneous albinism. Similarly, Noche et al[29] reported in 2010 that 5% of patients in schools for the visually impaired children in Yaoundé were diagnosed with low vision due to albinism.[29]

Developing countries also experience problems due to low socioeconomic standards. This may hinder accessibility to quality health care, especially among patients with low vision, and most especially among those with oculocutaneous albinism. This situation is further compounded by the fact that oculocutaneous albino patients have difficulties integrating into society. Low economic standards also hinder the acquisition of the up-to-date equipment needed for the optimal management of low vision.

This study is limited by the small sample size and the fact that autorefraction was used to determine the refrac-

Table 5 Mean ages of the patients with and without improvements in visual acuity

Evolution of VA	Mean CDVA	n	Mean age (y)	Standard deviation	95% CI	*P**
G1	0.20 ± 0.09	36	13.00	8.99	9.96–16.04	0.442
G2	0.11 ± 0.04	34	11.56	6.26	9.38–13.74	
Total	0.15 ± 0.08	70	12.30	7.76	10.41–14.13	

Notes: *Determined using Student's *t*-test. G1: distance VA was improved with optical correction. G2: distance VA could not be improved with optical correction.
Abbreviations: VA, visual acuity; CDVA, corrected distance visual acuity; CI, confidence interval; G1, group 1; G2, group 2; y, years

Table 6 Multivariate logistic regression modeling

Variable	Coefficient	Odds ratio	95% CI	P*
Diopters	–0.04	0.96	0.78–1.18	0.695
Age	0.03	1.03	0.96–1.11	0.384
Refractive error				
Myopia	Reference	Reference	Reference	Reference
MA	–1.35	0.26	0.06–1.17	0.049
Hypermetropia	0.43	1.54	0.22–11.04	0.666
HA	–2.27	0.10	0.01–0.76	0.026
Mixed astigmatism	–	–	–	–

Note: *The dependent variable was improvement in visual acuity.
Abbreviations: CI, confidence interval; MA, myopic astigmatism; HA, hypermetropic astigmatism.

tion statuses of our patients. Nystagmus is common in oculocutaneous albino patients and could have led to inaccurate measurements.

Conclusion

Complete oculocutaneous albinism is a condition that causes low vision. Myopic astigmatism and myopia were the most common refractive errors identified in this series. Optical correction is an important method for improving distance visual acuity in these patients.

Disclosure

The authors report no conflicts of interest in this work.

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