



# Refractive Profile in Oculocutaneous Albinism in Abidjan

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## Abstract

**Introduction:** Oculocutaneous albinism (OCA) is a rare genetic disorder affecting the production of melanin, with significant consequences for skin, dander and visual function. This article explores the clinical and therapeutic aspects of OCA, with an emphasis on associated refractive abnormalities. **Methods:** We conducted a bicentric retrospective study in two private pediatric ophthalmology referral centers in Abidjan over a period of two years, analyzing the records of patients with OCA seen for consultation. **Results:** Thirty-eight patients aged one to 23 years were enrolled. The prevalence of oculocutaneous albinism was 2.17%. The mean age was  $9.07 \pm 5.77$  years. The sex ratio was 2.1. The familial nature of OCA was reported in 57.9% of cases. Nystagmus was horizontal and pendular in the majority of cases (97.7%) and associated with strabismus in 28.9%. Refraction was pathological in 97.4% of cases. Astigmatism was found in all ametropic patients. This refractive defect was simple (5.4%) or associated with another ametropia, notably myopia (62.2%) or hyperopia (32.4%). Astigmatism was moderate in 64.9% of patients. Fourteen patients (36.8%) had anisometropia, which was myopic in 13.2%, hyperopic in 2.6% and astigmatic in 10.5%. All patients benefited from total optical correction and appropriate treatment of their corrective lenses. **Discussion:** OCA is a condition with a major visual and social impact. Early ophthalmological treatment is essential to limit the functional consequences, particularly amblyopia. Efforts must be made to improve access to care and visual aids, particularly in Africa where the prevalence is high.

## Subject Areas

Ophthalmology's Clinical Study

## Keywords

Oculocutaneous Albinism, Refraction, Anisometropia, Amblyopia

## 1. Introduction

Albinism is a universally inherited condition resulting from a defect in the synthesis of melanin, a pigment essential for skin protection and visual function. Oculocutaneous albinism (OCA) is the most common form of hereditary generalised hypopigmentation, which is inherited in an autosomal recessive fashion. There are several types, which have been classified on the basis of both the clinical forms and the genetic aspect, which is based on the genotype. AOC types 1 to 7 and syndromic forms are distinguished. Worldwide prevalence varies between 1/17,000 and 1/40,000 [1]; that of AOC type 3, which is more common in Africa, is 1/8500 [2]. In addition to the pigmentary defect, albino patients had significant ocular anomalies, in particular: nystagmus, strabismus and refractive defects causing visual impairment, which increased amblyopia even further in the absence of optical treatment [2]. In view of the paucity of local and sub-regional studies in West Africa on the problems associated with OCA, this study was initiated with the aim of identifying the refractive profile of oculocutaneous albinism in Côte d'Ivoire in order to provide the best possible care.

## 2. Method

A bi-centric study in private ophthalmology centers in Abidjan was carried out over a 24-month period from 01 January 2021 to 31 December 2022, consisting of a retrospective analysis of the records of patients with oculocutaneous albinism (OCA) seen for consultation.

The inclusion criteria were records of albino patients under 16 years of age who had undergone all stages of the clinical examination confirming the diagnosis of OCA. Incomplete records were not included, nor were medical records from children whose parents had refused to participate in the study.

A complete ophthalmological examination of all patients included a sensory assessment with measurement of distance visual acuity. For children of preverbal age, a behavioral method was used: fixation, eye-tracking and blinking on threat. For the stereoscopic vision test, we used the LANG and TNO test. A motor assessment was used to determine the presence of ocular deviation (phoria or tropia), specifying as far as possible the angle of deviation and the fixating eye. The type and characteristics of the nystagmus were studied. The refractive assessment included a subjective refraction using an automatic refractometer on a table (Topcon KR 800) or a portable refractometer of the Retinomax type (K+ Screen), followed by a systematic objective refraction under cycloplegia using atropine or cyclopentolate, depending on the child's age and cooperation.

The type of ametropia and its degree were specified. For the analysis of the sphere, we used the spherical equivalent (SE) considering a single eye (right eye). For the analysis of the cylinder, we used the basic cylinder. According to the dioptric power, low myopia was between  $-0.50$  and  $-2.75$  D, moderate between  $-3.00$  and  $-5.75$  D and high myopia was  $\geq -6$  D. Hyperopia was mild between  $+0.50$  and  $+2$  D, moderate between  $+2$  and  $+5$  D and strong if  $\geq +5$  D. Mild astig-

matism was between 0.75 and 2.00 D, moderate between 2 and 4 D and strong above 4 D. Anisometropia was defined as a dioptric difference of one diopter for the sphere and/or 0.75 D for the cylinder. The information was collected confidentially using an anonymous survey form in agreement with the managers of the two ophthalmology clinics. These ethical and deontological considerations are in accordance with the principles of the Declaration of Helsinki.

The data collected were processed and analysed using Word and Excel software and EPI INFO version 7.2. The limitations of this study lie in its retrospective nature, which makes it difficult to exploit certain information.

### 3. Results

Based on the selection criteria, 38 cases of oculocutaneous albinism were identified out of 1749 patients seen during the study period, representing a hospital prevalence of 2.17%. The majority of patients (55.3%) were aged between 6 and 15 years, with an average age of  $9.05 \pm 5.77$  years and extremes of one year and 23 years. The sex ratio was 2.1. Family factors and consanguinity were observed in 2.6% of cases.

The family nature of the AOC was notified in 57.9% of cases, and concerned 2nd and 3rd degree relationships in 44.7% and 5.3% of cases respectively. In the clinical study, the nystagmus was horizontal and pendular in the majority of cases (97.7%). Strabismus was associated with 28.9% of patients. All patients were ametropic (97.4%) except for one 3-year-old child who had physiological hyperopia. Astigmatism was found in all ametropic patients. This refractive defect was simple (5.4%) or associated with another ametropia, notably myopia in 62.2% or hyperopia in 32.4% (**Table 1**). Astigmatism was moderate in 64.9% of patients and mild in 27.7%. Mild myopia was observed in 39.13% of patients. It was moderate or severe in 26.09% and 34.78% respectively. The distribution of the degree hyperopia was almost identical for mild, moderate and high hyperopia (**Table 2**). Fourteen patients (36.8%) had anisometropia, which was myopic in 13.2%, hyperopic in 2.6% and astigmatic in 10.5% (**Table 3**). All patients received full optical correction.

**Table 1.** Distribution by type of ametropia.

Ametropia	Workforce	Percentage
Astigmatism	2	5.4
Hyperopia + Astigmatism	12	32.4
Myopia + Astigmatism	23	62.2
Total	37	100

**Table 2.** Distribution of patients by type and degree of refractive error.

Degree	Astigmatism	Hyperopia	Myopia
High	2 (5.4%)	5 (38.4%)	8 (34.78%)
Moderate	24 (64.9%)	4 (30.8%)	6 (26.09%)
Mild	11 (27.7%)	4 (30.8%)	9 (39.13%)
Total	37	13	23

**Table 3.** Distribution of patients according to anisometropia.

Anisometropia	Workforce	Percentage
Aniso-astigmatism	4	10.5
Aniso-hypermetropic	1	2.6
Aniso-myopia	5	13.2
Anisomyopia + anisoastigmatism	4	10.5
Absent	24	63.2
Total	38	100.0

#### 4. Discussion

The analysis of this study is based on data collected in Abidjan, highlighting the hospital prevalence, clinico-refractive profile and management of OCA in an African context. Hospital prevalence was 2.17% of all ophthalmological consultations. According to the literature, prevalence ranges from 1/20,000 to 1/35,000 cases in industrialised countries and 1/1000 to 1/5000 cases in sub-Saharan Africa [3]. According to the Ebana hospital study in Cameroon, the prevalence was 0.15% [4]. The high trend in our series can be explained by a selection bias. Indeed, patients with OCA are regularly, if not systematically, referred to our two departments, which are national reference centres for paediatric ophthalmology and strabology. Patients ranged in age from one to 23 years, with an average age of  $9.05 \pm 5.77$  years. In a study carried out in Burkina Faso, the author reported an average age of  $19.64 \pm 15.7$  years, ranging from 6 months to 55 years [5]. Our results, which differ from those of Konaté, could be explained by a methodological bias, as Konaté's series included all patients of all ages with OCA, whereas we collected a sample of young patients seen in the paediatric ophthalmology department. Males predominated in 68.4% of patients, with a sex ratio of 2.1. This result is identical to that of Gargouri [6] in Tunisia. Unlike X-linked ocular albinism, in which only males are affected and females are healthy carriers, OCA affects both sexes without preference. In family data, the notion of consanguinity was reported in only 2.6% of cases. This result is quite contrary to those of Ba [3] in Bamako with 37.2% of consanguineous union and Gargouri [6] in Tunisia, where consanguinity was observed in the entire study population. This can be explained by the fact that in these regions of Africa, marital union within siblings is still prevalent and linked to cultural factors. This consanguinity favours the occurrence of numerous genetic anomalies at the origin of pathologies such as AOC. A family history of albinism was reported in 57.9% of patients. Clinically, nystagmus was observed in almost all cases (97.4%). It was pendular and horizontal, as described by Aoubakar [7]. The mechanism responsible for nystagmus in albinism is not well understood. Studies suggest that abnormal development of the oculomotor system and abnormal organisation of the lateral geniculate bodies and their projection towards the cortex are determining factors in the appearance and persistence of nystagmus [8]. In short, nystagmus is very common in patients with

OCA. Strabismus was associated with nystagmus in 28.9% of patients. This result is lower than those of Ebana [4] and Aboubakar [7] in Cameroon, who found 33.33% and 52.8% respectively in their studies. The various constitutional ocular anomalies observed in albinism could explain this association. As strabismus is an amblyogenic factor, it leads to an additional visual handicap in patients with OCA. Refractive errors were present in 97.4% of cases, and only one patient had so-called physiological hypermetropia in relation to his age. The most common refractive error was astigmatism, which was present in all ametropic patients. This finding is consistent with the conclusions drawn by Eballé [9] and Aboubakar [7], and would be due to the presence of horizontal nystagmus in all these patients. Astigmatism associated with myopia was found in 60.5% of patients. This result corroborated that of Ebana [4] with 61.9%. Astigmatism associated with hyperopia was found in 31.6% of cases, whereas Ba and Eballé found 16.5% and 14.3% respectively [3] [9]. Astigmatism of low to moderate degree accounted for more than two thirds of our patients, in contrast to Eballé's series which reported a majority of astigmatism of high dioptric degree [9]. Concerning hyperopia, the distribution was homogeneous according to the different dioptric degrees at unlike Sayed [10] who found a preference for strong hyperopia. The prevalence of anisometropia in our study was 36.8%. This prevalence is still high compared with the data in the literature on anisometropia, which varies between 4.7% and 7.5% [11]. Aniso-myopia was the most frequent form with 13.2% of cases, followed by aniso-astigmatism (10.5%). The high prevalence of refractive anomalies highlights the importance of early detection and treatment. Even moderate anisometropia has major consequences for visual function, including amblyopia in the more anisometropic eye [12]. The main challenge of early detection of refractive disorders is to obtain the best possible visual acuity, as this will enable the child to become a future social player with unrestricted access to the labour market. The associated visual impairment will lead to relational difficulties and poor sensorimotor experiences [13]. All our patients have benefited from total optical correction. This practice should be encouraged to ensure optimal vision and improve quality of life. All patients with oculocutaneous albinism should wear a total optical correction obtained after measurement of objective refraction under cycloplegia. According to some authors, even if there is no curative treatment for OCA, it is necessary to prescribe suitable corrective lenses (filtering or tinted lenses) or the use of rigid lenses in cases of significant astigmatism [2]. Psychological support and educational assistance are also essential to help albino patients integrate into society.

### Conflicts of Interest

The authors declare no conflicts of interest.

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