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Ocular Manifestations Encountered in Albinos Living in Libreville: Epidemiological and Clinical Aspects

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Abstract

Introduction: Albinism is a rare hereditary genetic disorder, characterized by melanogenesis disorders associated to varying degrees of cutaneous, atrial and visual disorders. In Libreville, no data on the subject is available. Purpose: To describe the ocular manifestations found in albinos living in Libreville. Patients and methods: This was a cross-sectional and descriptive study that took place in Libreville during a year from and that concerned 43 albinos. The inclusion criteria were any albino with oculocutaneous albinism present in Libreville who has agreed to participate in the investigation. The variables studied were age, sex, visual acuity, ametropia, iris color, iris transillumination, strabismus, nystagmus, torticollis, amblyopia, optic disc, photophobia, foveal hypoplasia and retinal hypopigmentation. Data was collected and analyzed using Epi infoTM 7.2.0.1 CDC and IBM® SPSS® V21 Statistic software. Results: The mean age was 21.2 ± 17 years with a (F/M) ratio of 0.53. Visual acuity by far without correction (AVLC_{SC}) < 3/10 was found in 88.4% of cases. Astigmatism, two-tone iris and torticollis were found respectively in 52.7%, 51% and 26% of cases. Retinal hypopigmentation and foveal hypoplasia were noted in 65.1% and 86% of cases. Divergent strabismus was noted in 71.4% and small and pale papilla in 95.1% of cases. Photophobia, iris transillumination, nystagmus and amblyopia were noted in 100% of cases. Conclusion: The ocular manifestations encountered in our series are similar to those described in literatures.

Keywords

Albinism, Nystagmus, Strabismus, Amblyopia, Torticollis, Photophobia, Libreville

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1. Introduction

Albinism is a group of hereditary disorders linked to a melanin biosynthesis abnormality, a pigment developed in the specialized cells of the skin, hair, iris, pigmented epithelium of the retina, the skin and internal ear, associated with a number and normal structure of melanocytes [1]. Albinism is a universal condition, the most common hereditary generalized hypopigmentation whose global incidence in all forms varies from 1/17,000 to 1/20,000 births [2] [3]. This is a clinically and genetically heterogeneous pathology with three major forms including Oculocutaneous albinism, Ocular albinism and Syndromic albinism. This condition varies from one form to another and from one continent to another with oculocutaneous albinism type 2 (OCA2) recognized as the most common form worldwide [1]. In America, its frequency for the entire population is about 1/36,000 compared to that of African-Americans with an incidence of 1/12,000 [4] and the Africans especially in Niger where it can reach 1/1000 [5]. Albinism Oculocutaneous type 1 (OCA1) uncommon among African blacks and African-Americans is more common among Caucasians. The type of Oculocutaneous albinism type 3 (OCA3) has mainly been described in Black-African and African-American with two cases reported among Caucasians. However, Oculocutaneous albinism type 4 (OCA4) remains the most common form of tyrosinase positive OCA in Japan [5]. In sub-Saharan Africa, in studies conducted in Cameroon, the hospital prevalence of albinism Oculocutaneous type 2 is 0, 15% according to Ebana [6] and 1.15% according to Kassir [7]. However, despite the explanations of science, albinism remains an anomaly source of fear and anxiety in Africa where albinos, victims of many prejudices are discriminated against, marginalized, forced to drop out of school and suffer physical and moral violence or even murdered for their organs, which are sold at high prices. As a result, they are confronted with a whole range of problems that constitute a hindrance to their personal growth, their school attendance, their social and professional integration and their medical care. Despite its frequency and the importance of the medical problems encountered, ocular involvement related to albinism, which is constant and serious, remains unknown in the albino subject living in Libreville in view of the only study made in 1872 [8]. Hence the initiation of this preliminary work in order to study the ocular manifestations encountered in albinos living in Libreville.

2. Patients and Method

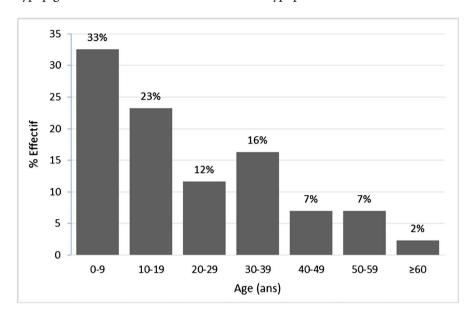
This was a descriptive cross-sectional study that took place in Libreville in the Ophthalmology Department of the Angondjé University Hospital and within a private structure of the place during the period of February 1, 2017 to February 1, 2018. The study population consisted of 43 albinos with Oculocutaneous albinism identified after albino sensitization in the albino welfare association (ALBA). The inclusion criteria concerned all albinos with oculocutaneous albinism living in Libreville and having agreed to participate in the study. Exclusion

criteria were imposed on albinos with no oculocutaneous albinism or who did not live in Libreville or who refused to participate in the survery. Consultation of each albino was made after albumin sensitization by the ALBA association. The patients listed were seen in a defined order of passage based on their availability and enthusiasm to participate in the survey. They were examined free of charge by the same medical team after obtaining their informed consent. The variables studied were age, sex, visual acuity, ametropia, iris color, iris transillumination, strabismus, nystagmus, torticollis, amblyopia, optic disc, photophobia, foveal hypoplasia and retinal hypopigmentation. All collected information was collected using a data collection sheet. The purpose of the interrogation was to obtain information on the patient's age, age of onset of ocular manifestations, and sex. The study helped to look for a torticollis by monitoring the fixation and supplemented or completed by the study of the oculomotricity in search of a strabismus and nystagmus. The ophthalmological examination, which was adapted to the patient's age, first included the study of visual acuity for each eye, then binocular, with and without correction. The visual acuity from a distance was made at a distance of 5 meters in the standard consultation room with the Monoyer Chart and the Snellen's E eye chart. The patients with visual acuity between 8/10 and 10/10 were considered emmetropic and those with visual acuity below 8/10 ametropic. Amblyopia was defined for any visual acuity less than 3/10. Close visual acuity was assessed using the Parinaud or Rossano Weiss reading scale placed 33 cm from the patient, which was considered good between P10 and P3 and bad if less than P3. All the ametropic patients benefited from an objective refraction with the aid of an automatic refractometer after instillation of a cycloplegic essentially atropine eye drops in our series according to two protocols. In children according to the age, we instilled in both eyes a drop of atropine 1%, 0.5% or 0.3% morning and evening for 7 days and in adults, a drop of tropicamide alternately every five minutes with a drop of cyclopentolate for 25 minutes. Biomicroscopy has allowed inter alia the study of iris coloration, photophobia and iris transillumination. The iris coloration was perfectly observable through the transparent cornea. However, the study of iris transillumination had required examination of the slit lamp by the application of a light source on the sclera or lower eyelid. When found, it was classified into four stages according to the morphological classification established by Dr. B. Kässmann-Kellner. Photophobia was also studied by looking at a light source. The detection of nystagmus was done in monocular and binocular vision by the fixation of an optotype in the different directions of the gaze and in convergence. It made it possible to highlight its characteristics, in particular its morphology, its direction and its intensity. Depending on the intensity, the nystagmus was weak if it was visible only with the slit lamp, then the nystagmus was so visible to the naked eye at a distance less than or equal to 3 m and finally the nystagmus was strong if we could see it beyond 3 m. The strabismus was objectified by the motor assessment which allowed the study of the deviation of the angle by the

method of corneal reflections or reflections of Hirshberg. The patient fixing a point of light in front of him to 30 cm and with an ophthalmoscope, we study the direction of the corneal reflection. When the reflection was centered, there was no strabismus. On the other hand, when the reflection was nasal, we had a divergent strabismus and when it was temporal, strabismus was convergent. Lastly, strabismus was vertical when the reflection was higher or lower. The intraocular pressure (IOP) measured using the GoldmanTM aplanation tonometer was considered normal for values between 10 and 21 mmHg. Posterior segment examination using the ophthalmoscope after dilatation by mydriatic allowed the study of the papilla, the fovea and the retina. Papillary and retinal abnormalities for hypopigmentation were classified according to B. Kasmann's classification. The foveal reflection when present gave a normal appearance of the fovea and when absent the fovea was considered hypoplastic. All these data collected on a survey sheet were reported in an Excel file of Microsoft Office 2010. Then, the analysis was done using the software Epi infoTM 7.2.0.1 CDC (Center for Disease Control and Prevention) and IBM® SPSS® Statistic 21. It used standard descriptive analyses (mean ± standard deviation). The Chi-2 test was used for the comparison of the proportions. The correlation was considered statistically significant for values of p < 0.05. This work was done according to the recommendations of the Helsinki Declaration of Ethics on the Use of Living Things. We obtained authorizations from the managers of the structures in which we conducted our study. Informed consent was obtained from the participants. In addition, we assured them of the confidentiality of the data collected.

3. Results

Our study population consisted of 43 albinos, 86 eyes. The mean age was 21.2 \pm 17.0 years with extremes of 8 month and 62 years and the most represented age group was 0 to 9 years in 32.6% of cases (Figure 1). Our study population consisted of 43 albinos including 15 males (34.9%) and 28 females (65.1%) a ratio of 0.53. Visual acuity by far without correction to 2 eyes (AVL_{SC}) < 3/10 was noted in 88.4% of cases. It was between 3/10 and 7/10 without correction to 2 eyes in 11.6% of cases (Table 1). Visual acuity by far with correction to 2 eyes (AVL_{SC}) < 3/10 was noted in 73.3% of cases (Table 2). Astigmatism was the most common ametropia with 52.7% of cases in both eyes, followed by myopia in 31.1% of cases and hyperopia in 16.2% of cases in both eyes (Table 3). Amblyopia was moderate in 84.8% of cases in both eyes and deep in 11.6% of cases in both eyes (Table 4). Iris had two tones in 70% of cases. It was blue in 2% of cases, brown in 23% of cases and gray in 5 of cases. Nystagmus, iris transillumination and photophobia were noted respectively in 100% of cases. Divergence strabismus was found in 71.4% of cases followed by convergent strabismus in 19% of cases. Vertical and mixed strabismus was observed in 4.8 cases respectively (Table 5). Torticollis was found in 26% of the albinos against an absence noted in 73% of the cases. Optic disc or papilla was small and pale in 81.3% of cases with retinal



hypopigmentation found in 65.1% and foveal hypoplasia in 86% of cases.

Figure 1. Distribution of albinism subjects by age group.

Table 1. Visual acuity without correction.

Visual Acuity Without Correction	Right Eye n (%)	Left Eye n (%)	2 Eyes n (%)
8 - 10	0 (0)	0 (0)	0 (0)
7 - 3	5 (11.6)	5 (11.6)	10(11.6)
<3	38 (88.4)	38 (88.4)	76 (88.4)
TOTAL	43 (100)	43 (100)	86 (100)

Table 2. Visual acuity with correction.

Visual Acuity With Correction	Right Eye n (%)	Left Eye n (%)	2 Eyes n (%)
8 - 10	0 (0)	0 (0)	0 (0)
7 - 3	10 (23.3)	13 (30.2)	23 (26.7)
<3	33 (76.7)	30 (69.8)	63 (73.3)
TOTAL	43 (100)	43 (100)	86 (100)

Table 3. Ametropia.

Ametropia	Right Eye n (%)	Left Eye n (%)	2 Eyes n (%)
Hyperopia	6 (15.8)	6 (16.7)	12 (16.2)
Myopia	12 (31.6)	11 (30.6)	23 (31.1)
Astigmatism	20 (52.6)	19 (52.7)	39 (52.7)
TOTAL	38 (100)	36 (100)	74 (100)

Table 4. Amblyopia.

Amblyopia	Right Eye n (%)	Left Eye n (%)	2 Eyes n (%)
Moderate	25 (89.3)	13 (86.7)	38 (88.4)
Deep	3 (10.7)	2 (13.3)	5 (11.6)
TOTAL	28 (100)	15 (100)	43 (100)

Table 5. Strabismus.

Strabismus	Right Eye n (%)	Left Eye n (%)	2 Eyes n (%)
Convergent	2 (14.3)	2 (28.6)	4 (19)
Divergent	11 (78.6)	4 (57.1)	15 (71.4)
Vertical	1 (7.1)	0 (0)	1 (4.8)
Mixed	0 (0)	1 (14.3)	1 (4.8)
TOTAL	14 (100)	7 (100)	21 (100)

4. Discussion

Limitations of the study

This survey encountered several limitations, the main one being the low participation or refusal of participation of many albinos to the study despite multiple phone calls, the invitation during awareness' meetings, free ophthalmological consultations, transportation and the rapid registration for obtaining health insurance. A number of reasons based on socio-cultural beliefs that albinos would be subject to human sacrifice may explain the finding already made by some authors such as Gassama in Mali and El-Ballouti in Morocco [9] [10].

Prevalence

The study population consisted of 43 albinos or 86 eyes. The prevalence of albinism in our series was 4.24%. This figure seems higher than the hospital prevalence reported by Ebana and Kassir in Cameroon in 0.15% and 1.15% respectively [6] [7] but remains not comparable to the figures cited because of the difference in the conditions the series was done. Indeed, our study took place at the end of an awareness period of the members of the association ALBA which indicates the non-promptness of the consultations but rather consultations motivated by being cost-free, by telephone calls, by paid transport, by the health insurance and others. The association ALBA currently has about 105 adherent albinos, if this population is related to the 850,000 inhabitants which counts Libreville, we can then estimate the prevalence of albinism in Libreville with 1.23/10,000 births approximately. According to studies, the prevalence of albinism varies from one country to another and from one region to another [11] [12]. This result differs from the prevalence found by Madelain with 1/20,000 and Risse with 1/15,000 [13] [14]. However, the prevalence of our series is currently unusable given the lack of epidemiological data on albinism at the national level.

Age

The average age of the population was 21.2 years with age boundaries of 8 months and 62 years. This figure is similar to that of Aboubakar [15] in Cameroon with 23.89 years and differs from those of Ajose [16] in Nigeria with 24.6 years, Gargouri [17] in Tunisia with 18 years, Ebana [6] in Cameroon with 13.11 years and Mokaya [18] in Kenya with 12.56 ± 4.16 years. However, this result reflects the youthfulness of the Libreville albino population which is similar to that of Aboubakar [15] in Cameroon. Nevertheless, this age remains late concerning the diagnosis and management of amblyopia, which must be done very early [6] [15]. According to studies, the life expectancy of albinos remains similar to that of the national population, but that of albinos is reduced to around 30 years because of the sacrifices and rituals they are subjected to in certain regions [9] [10]. Sex

The female population was the most represented in 65.1% of cases against 34.9% for men, a ratio of 0.5. This result corroborates those of Aboubakar and Mokaya with respectively a sex ratio of 0.5 and 0.7 [15] [18]. On the other hand, these figures differ from those of Ebana [6], Kassir [7], Gargouri [17], and Ajose [16] who found a male-dominated ratio in respectively 1.21; 1.29; 2.1; and 1.1. Indeed, these controversial results do not necessarily prove that albinism is a sex-related genetic disorder, but these figures can probably be justified by the strong tendency of women to participate in actions and associative life. However, this result is currently unusable because no figures were found at the national level.

The ocular manifestations

Photophobia

Photophobia is the main functional sign found in 100% of cases. This figure matches that of Mokaya [18] with 91% of cases and Ebana [6] with 100% of cases. According to the literature, photophobia is a constant symptom regardless of the regions. It can sometimes be disabling and nonspecific to Ocular cutaneous albinism because it exists in other retinal pathologies [2] and results from the reduction of secondary light filtration to melanin deficiency in all cases.

Visual acuity

In our series, the best visual acuity after correction was less than 3/10 in 73.3% of cases. This result corroborates the better visual acuity from afar after correction found by Ebana [6] and Aboubakar [15] in Cameroon and by Mokaya [18] in Kenya. Indeed, poor visual acuity is one of the main signs associated with albinism that is present at birth and ranges from 20/40 to 20/400 births in some cases [19] [20]. According to studies, albinism is responsible for 5% to 10% of the world's low vision and this poor visual acuity related to albinism is difficult and almost impossible to improve in the majority of cases in the albino subject as reported by several authors [6] [14] [15]. However, near vision was better in our study and albino cases could see up to P2 without correction in 45.2% and

with correction in 74.1% of cases. These results are consistent with those of other series who claim that despite the severity of low vision, near vision remains intact [21].

Ametropia

Astigmatism was the most frequently encountered ametropia in 52.7% of cases. This result represents half of Gargouri [17] with 100% of cases and of Aboubakar [15] in almost all cases. Indeed, according to studies, ametropia is constant in albinism and the most frequently encountered ametropia are strong astigmatism, medium or high hyperopia, however strong myopia is rare [2].

Strabismus

Strabismus was divergent in 71.4% of cases; this figure is different from that of Aboubakar [15] who noted it in 16.7% of cases. Indeed, according to studies, the strabismus encountered in albinism can be explained by abnormalities in the distribution of retino-geniculate and geniculo-cortical visual fibers that hinder the development of binocular vision [6] [14].

Nystagmus

Patients had congenital onset nystagmus in 100% of cases. This finding is also made by Ebana, Aboubakar and Mokaya [6] [5] [18] who had found a nystagmus in almost 100% of cases in their series. According to the literature, nystagmus in albinism is almost constant and can be discovered in the first months of life when it is full. It increases with glare and decreases with the use of optical correction [2]. This nystagmus is due to foveal hypoplasia and other ocular abnormalities frequently encountered in albinism [22] [23].

Torticollis

Torticollis in primary fixation by far was noted in 26% of cases in our series. This figure deviates from that of Ebana, which reported it in 35.7% of cases [6]. According to studies, most people with albinism adopt a compensatory head posture that reduces nystagmus and improve vision [2]. The morphological variability of the nystagmus describes different types namely horizontal torticollis, vertical or mixed [2] [21].

Iris transillumination

Iris transillumination was found in all our patients. This finding is also reported by Mokaya and Ebana, who reported iris transillumination in 87% and 100% of cases respectively [6] [18]. Indeed, due to melanin deficiency in the stroma and posterior epithelium of the iris, the light reflected by the retina is not filtered and patients with albinism then undergo iris transillumination. The latter can be complete, it is the classic pink eye, not obvious with, in this case a red pupillary reflection visible under certain incidences and sometimes incomplete, only visible to the slit lamp [24].

Amblyopia

Patients had moderate amblyopia in 88.4% of cases. Indeed, studies report that albinism is responsible for 5% to 10% of the world's low vision [2] [21]. However, amblyopia in the albino subject may be of functional or organic origin

and the risk of strabismus amblyopia in albinism is low [2] [21].

Foveal hypoplasia

Foveal hypoplasia was described in 86% of cases. This result tends towards that of Mokaya [18] which found it in 100% of the cases. According to studies, patients with albinism have characteristic anatomical deficits such as foveal hypoplasia and the abnormal decussation of the optic fibers [2] [21]. Foveal hypoplasia is probably the most significant limiting factor in albino vision and is thought to be due to a low amount of melanin in the retinal pigment epithelium [25] [26].

Retinal hypopigmentation

Retinal hypopigmentation was found in 65.1% of the cases in this series, this result being similar to that of Mokaya [18] who found it in 97.5% of cases. The poor vision, seen in albinos, is not only due to an absence of pigments that would prevent the formation of a dark room as in a normal eye. Histology shows an atypical retina, containing an abnormally poor fovea in cones. The retina is a neural tissue and the cone and rod receptor cells are in contact with the retinal pigment epithelium [10] [27].

Aspect of the optic disc

The papilla was small and pale in 81.3% of cases. Papillary morphological abnormalities are frequent and correlated with visual acuity level in albinism. These abnormalities were classified into four stages according to B. Kässmann, in a study of a series of more than 600 patients [28].

5. Conclusion

The ocular manifestations found in albinos living in Libreville are similar to those described in studies. They require ophthalmological follow-up or medical and surgical management if necessary. Photophobia exacerbated by astigmatism associated with amblyopia constitutes a real visual handicap by their constant and disabling character for albinos. Albino's refusal to adhere to organized health actions with a retreat on their own would be due to the different mystical-political beliefs encountered in Africa and elsewhere. It would be desirable for the policies to take this statement into account and consider it a real public health problem to facilitate the social and professional integration of all albinos living in Libreville.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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