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Refractive State and Macular Abnormalities at Oct in Albino at Chu Iota

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Abstract

Introduction: A rare genetic disease, albinism is globally characterized by specific deficits of the visual system associated with a variable hypopigmentation phenotype depending on the disruption of melanin production [1]. It is linked to a hereditary defect in the biosynthesis of melanin. Disease results in a generalized decrease in the pigmentation of the appendages, skin and eyes [2]. The aim of this study is to determine the different refractive errors and the different macular anomalies during the OCT examination in oculocutaneous albinism at the CHU-IOTA. **Patients and Method:** We conducted a prospective study in patients over 10 years of age with oculocutaneous or ocular albinism who consulted at the CHU-IOTA between July 1, 2020, and September 30, 2021. **Results:** Over the period, 105 cases of oculocutaneous albinism were collected out of a total of 42,024 consultations, which corresponds to a frequency of 0.2%. The average age was 26.2 years, (11 years to 48 years). The sex ratio was 1.6. Astigmatism was the most found refractive error in 50.48% of cases, followed by myopia in 29.52% of cases and farsightedness in 20% of cases. The macular thickness between 251 - 350 was the most commonly found in both eyes, *i.e.* 47.25% on the right and 53.55% on the left. The bulging macula was the most frequent pathology on the OCT at the level of the two eyes, *i.e.* 41.42% on the right and 50.6% on the left. **Conclusion:** Following the visual impairments linked to albinism, early optical care and access to OCT are necessary. Thus the accompaniment of a subject with albinism and associations of albinism must be global and meet specific needs, in order to prevent or avoid ocular complications.

Keywords

Refractive Defects, Macula, OCT, Albinos, CHU-IOTA

1. Introduction

A rare genetic disease, albinism is characterized globally by specific deficits of the visual system associated with a variable hypopigmentation phenotype depending on the disruption of melanin production [1]. It is linked to a hereditary defect in the biosynthesis of melanin. Disease results in a generalized decrease in the pigmentation of the appendages, skin and eyes [2]. Despite the fairly advanced knowledge of the genetics of albinism, approximately 20% of affected patients currently remain without a genetic diagnosis [3]. Poor visual acuity is one of the main signs of albinism. This visual impairment is more often moderate but can be profound [4]. Ametropia, nystagmus, strabismus, photophobia and macular abnormalities including foveolar hypoplasia are the cause of visual difficulties variously associated with albinism and produce varied clinical pictures [5]. It is not specific during photophobia and oculomotor signs [6]. There are several forms of albinism including oculocutaneous albinism classified into four main forms, three minor forms and one form of ocular albinism [1] [7]. And the prevalence of all forms of albinism varies widely around the world and has been estimated at around 1/17,000 [1]. It would be the cause of 5% to 10% of visual impairment in the world [8]. Very few studies have been conducted, hence the interest of our work at the CHU-IOTA in order to determine the various refractive anomalies and OCT data in macular anomalies in albinos.

2. Patients and Method

We conducted a prospective study between July 1, 2020, and September 30, 2021, *i.e.* a period of 15 months. The study included any subject over 10 years old with oculocutaneous or ocular albinism who consented to participate in the study.

The subjects were contacted by telephone and invited to the CHU IOTA, they had explanations of the objectives of the study and the procedure of the examination.

All subjects underwent an ophthalmological examination including measurement of visual acuity, objective refraction, biomicroscopic examination of the anterior segment, fund us examination and macular OCT.

Classification of albinism was based on hair color at birth and after birth, skin color and iris color [4] and The grade of foveolar hypoplasia was defined by the classification of Thomas *et al.* in 4 grades: absence of extrusion of the plexiform layers (grade 1), absence of a foveolar depression (grade 2), widening of the outer nuclear layer (grade 3), elongation of the outer segments (grade 4). [9].

The data studied were age, sex, type of oculocutaneous albinism, VA before correction, VA after correction, refractive values, macular thickness, and macular anomalies. The data was collected through a survey form and a medical file developed for this purpose. Data collection and analysis were done using IBM SPSS 20.0 software. Word processing and tables were carried out by Microsoft Office software version 2016.

3. Results

Over the period, 105 cases of oculocutaneous albinism were collected out of a total of 42,024 consultations, *i.e.* a frequency of 0.2%. There was a male predominance in the study population with an M/F sex ratio equal to 1.6. The 21 - 40 age group was the most represented at 49.2% (Table 1). Consanguinity was noted in 99.05% of cases and 89.52% of patients had family members with albinism (Table 2). The most represented VA without correction was between 1/10-3/10 in both eyes, *i.e.* 46.6% on the right and 50.5% of cases on the left (Table 3). The most found VA with correction was also between 1/10-3/10, *i.e.* respectively 59.05% in the right eye and 57.14% in the left eye (Table 3). We recorded 13.33% and 11.43% of functional amblyopia on the right and left respectively (Table 3).

4. Discussion

In albinism, all ametropias can be found. The optical correction of these refractive errors is the initial management of albino patients, it can be done by optical glasses or by contact lenses at best [10] [11]. This optical correction must be early from a young age with an annual reassessment [7]. During our study we also found all forms of ametropia with a predominance of astigmatism at 50.48%, the mean was −3.50 D; followed by myopia which represented 29.52% with an average of −8 D; farsightedness accounted for 20% with an average of +3.50 D. All our patients received optical correction and the majorities were already wearing glasses. Astigmatism is usually the most common and is often

Table 1. Distribution of patients by history.

Inbreeding	Number	Percentage
Yes	**104**	**99.05**
No	1	0.95
Total	105	**100.00**
Familial albinism	Number	Percentage
Yes	**94**	**89.52**
No	11	10.48
Total	105	100.00

Table 2. Distribution of patients according to distance VA ODG without correction.

	OD		OG	
	Number	Percentage (%)	Number	Percentage (%)
<1/10	30	28.6	30	28.6
[1/10 - 3/10[	**49**	**46.6**	**53**	**50.5**
>3/10	26	24.8	22	20.9
Total	105	100	105	100

Table 3. Distribution of patients by distance VA ODG WITH correction.

	OD		OG	
	Number	Percentage (%)	Number	Percentage (%)
<1/10	14	13.33	12	11.43
[1/10 - 3/10[	**62**	**59.05**	**60**	**57.14**
>3/10	29	27.62	33	31.43
Total	105	100	105	100

associated with hypermetropia hyperopia or myopia [11]. S. Gargouri *et al.* found 59% farsightedness, 41% myopia and 100% astigmatism with also 100% consanguinity [11]. The most common universal condition of hereditary generalized hypopigmentation, oculocutaneous albinism type 2 is the most common form in the world as well as in black Africa [1]. It represented more than half of the cases in the study, *i.e.* 51.45%. Protection against light is essential for albinos, especially in sunny countries. It reduces associated symptoms including glare, photophobia and nystagmus due to excessive stimulation of the hypopigmented retina by light and therefore improves vision [5]. Visual improvement is significant in myopes, especially at low diopters but this is variable [10]. We then obtained 87.15% visual improvement and 23.1% functional amblyopia.

These cases of amblyopia could be explained by the delay in the initial management, in particular of photophobia, nystagmus and high myopia by optical correction [12]. Most of the improvements were achieved in myopic patients between −2.50 and −8.50 D. In Schulza S the greatest improvement was achieved in myopic patients between −1 and −7.50 D [10]. Foveolar hypoplasia is the most common macular involvement in oculocutaneous albinism, even if it is not pathognomonic. It is visible on fundus examination and easily found on OCT examination [9]. Macular optical coherence tomography (OCT) was performed in all our patients. Grade 2 hypoplasia was the most frequent macular anomaly with 41.42 on the right and 50.6% on the left, followed by grade 4 with 31.61 on the right and 34.5% on the left, followed respectively by widening of the outer nuclear layer (grade 3) with 31.61% on the right and 34.5% on the left, elongation of the outer segments (grade 4) on the right 14.17% and 23% on the left and absence of extrusion of the plexiform layers (grade I) on the right 6.54% and on the left 6.9%. We also noted less than 5% of cases of retinoschisis and more than 6% of pachychoroid, and the macula was normal in 14.17% on the right and 5.75% on the left.

Grade 2 hypoplasia, a rare complication of high myopia, corresponds to a convex protrusion of the macula within a myopic staphyloma facing an area of scleral thickening [13]. Given the high frequency of myopia in our study, this could be the case with our patients.

5. Conclusion

A rare and universal genetic disease, albinism is accompanied by ametropia,

nystagmus, strabismus, photophobia and macular abnormalities including foveolar hypoplasia. Hence, the visual impairments are in cases of oculocutaneous albinism. Their detections are based on refraction and macular OCT. Early management of these fraction defects through optical correction is necessary to avoid functional amblyopia.

Conflicts of Interest

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

References

[1] Mártinez-García, M. and Montoliu, L. (2013) Albinism in Europe. *The Journal of Dermatology*, **40**, 319-324. https://doi.org/10.1111/1346-8138.12170

[2] Sauer, A. and Speeg-Schatz, C. (2012) Albinisme ocul-cutanée. *Journal Français d'Ophtalmologie*, **35**, 851. https://doi.org/10.1016/j.jfo.2012.01.004

[3] Montoliu, L., Grønskov, K., Wei, A.H., Martínez-García, M., Fernández, A., Arveiler, B., Morice-Picard, F., Riazuddin, S., Suzuki, T., Ahmed, Z.M., Rosenberg, T. and Li, W. (2014) Increasing the Complexity: New Genes and New Types of Albinism. *Pigment Cell & Melanoma Research*, **27**, 11-18. https://doi.org/10.1111/pcmr.12167

[4] Rooryck, C., Morice, F., Mortemousque, B., Lacombe, D., Taïeb, A. and Arveiler, B. (2007) Albinisme oculo-cutanée. *Annales de Dermatologie et de Vénéréologie*, **134**, 55-64. https://doi.org/10.1016/S0151-9638(07)89250-7

[5] Omar, R., Idris, S.S., Meng, C.K. and Knight, V.F. (2012) Management of Visual Disturbances in Albinism: A Case Report. *Journal of Medical Case Reports*, **6**, Article No. 316. https://doi.org/10.1186/1752-1947-6-316

[6] McCafferty, B.K., Wilk, M.A. and McAllister, J.T. (2015) Clinical Insights into Foveal Morphology in Albinism. *Journal of Pediatric Ophthalmology & Strabismus*, **52**, 167-172. https://doi.org/10.3928/01913913-20150427-06

[7] Le Meur, G. (2017) Pathologie rétinienne et dégénérative et/ou héréditaire (Albinisme). Rapport SFO-Ophtalmologie pédiatrique.

[8] Savananthan, N., Surendran, M. and Roberts, E.O., Jain, S., Thomas, S., Shah, N., Proudlock, F.A., Thompson, J.R., McLean, R.J., Degg, C., Woodruff, G. and Gottlob, I. (2009) The Prevalence of Nystagmus: The Leicestershire Nystagmus Survey. *Investigative Ophthalmology & Visual Science*, **50**, 5201-5206. https://doi.org/10.1167/iovs.09-3486

[9] Thomas, M.G., Kumar, A., Mohammad, S., Proudlock, F.A., Engle, E.C., Andrews, C., Chan, W.M., Thomas, S. and Gottlob, I. (2011) Structural Grading of Foveal Hypoplasia Using Spectral-Domain Optical Coherence Tomography a Predictor of Visual Acuity? *Ophthalmology*, **118**, 1653-1660. https://doi.org/10.1016/j.ophtha.2011.01.028

[10] Schulze, S.M., Kumar, N. and Bohrmann, D., Msukwa, G., Kalua, K., Kayange, P. and Spitzer, M.S. (2015) Refractive Errors, Visual Impairment, and the Use of Low-Vision Devices in Albinism in Malawi. *Graefe's Archive for Clinical and Experimental Ophthalmology*, **253**, 655-661. https://doi.org/10.1007/s00417-015-2943-0

[11] Gargouri, S., Kaibi, L., Sakka, M., Abib, L., Sallami, D., *et al.* (2017) La prise en

charge optique des albinos en Tunisie. *Journal de l'Information Médicale de Sfax N°* 25, **17**, 40-44.

[12] Khanala, S., Pokharelb, A. and Kandel, H. (2016) Visual Deficits in Nepalese Patients with Oculocutaneous Albinism. *Journal of Optometry*, **9**, 102-109. https://doi.org/10.1016/j.optom.2015.01.002

[13] Gaucher, D. and Leveziel, N. (2019) Macula bombée du myope fort. Rapport SFO.