

Visual Defect in Oculocutaneous Albinism is Not Associated with Gross Structural Anomaly

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Abstract

Background: Albinism is a heterogeneous group of inherited non progressive disorders of melanin metabolism. The two main types are Ocular albinism (OA) in which pigment is absent only in the eyes and Oculocutaneous albinism (OCA) in which the eyes, skin and hair lack pigment. The tropical environment, without the protective effect of melanin, predisposes the African OCA to skin cancers. In the eyes fovea maturity is impaired leading to poor vision. All forms of albinism, regardless of the genotype or phenotype, have the same distinctive visual impairment that confers visual acuity ranging from 20/40(6/12) to 20/200(6/60) that significantly limits their education, occupation and recreation. This study set out to use ultrasonography to detect correctable ocular structural anomaly in visually impaired African OCA who also have severe sun-damaged skin.

Materials and methods: In a prospective study, the eyes of 57 consenting Nigerian OCA referred from Dermatology to Radiology for ocular scan were investigated with B-mode ultra-sonogram. The results were compared with matched controls and analyzed by simple descriptive statistics.

Results: The age range of the study population was 15 to 62 years (mean 24.6 years) and male: female ratio was 1:2. Of the OCA, 98% and of the controls, 91.2% had normal ocular scans. Vitreous echoes were found in 7.0% of all participants and one (1.8%) of the control participants had cataract. No cataract or other gross structural anomaly was detected in any of the orbital structures of the OCA.

Conclusion: Ultrasound scan reveals no ocular structural abnormalities peculiar to OCA. Though not statistically significant, there is an intriguing absence of cataract in OCA that is worthy of further investigation.

Keywords: African; Oculocutaneous albinism; Ocular scan; Cataract

Introduction

Albinism is a heterogeneous group of inherited non progressive disorders of melanin metabolism manifesting a wide variety of phenotypes, limited number of genotypes and a rather complex genetics [1]. The primary abnormality is a mutation of one of several genes that code the instruction for making one of the many proteins necessary for melanin production. The main function of the melanin pigment includes the filtering of ultraviolet radiation (UVR) entering the skin to prevent sunburn and skin cancer. The fovea of the eye cannot reach full development in the absence of melanin [2].

The two main types are Ocular albinism (OA) which affects only the eyes and Oculocutaneous albinism (OCA) which affects the eyes, skin and hair. In OCA there is little or no pigment in the eyes, skin and hair, while OA lacks pigment in the eyes only with skin and hair colour varying from very fair to dark [1].

The three subtypes of OCA found in Africans; OCA 1, OCA2 and Brown OCA (BOCA) are all autosomal recessive disorders that manifest either a complete lack or a reduction in melanin pigmentation of the hair, skin and eyes [3]. Their features overlap and are more accurately distinguished by genetic testing. Regardless of the genotype or phenotype, all forms of albinism have the same distinctive visual impairment with varying severity. Ophthalmologic findings comprise misrouting of the optic nerve fibre radiations at the chiasm resulting in strabismus, reduced stereoscopic vision and distinctive visually evoked potentials (VEP) [1,4,5]. Other components are photophobia, nystagmus from infancy, hypopigmentation of the iris and fundus. Foveal hypoplasia leads to reduced visual acuity that range from 20/40(6/12) up to 20/200(6/60) [1,4,5].

Albinism affects people of all ethnic backgrounds. Prevalence varies

considerably worldwide and has been estimated at approximately 1: 17,000 to 1: 20,000. In Nigeria it is considered to be 1:15,000 [5,6].

The tropical environment, without the protective effect of melanin, predisposes the African OCA to the harmful effects of ultraviolet radiation. The skin develops sunburns, blisters, solar keratoses, lentigines, ulcers and eventually multiple aggressive skin cancers from which they may die in early adult life or middle age [6,7]. Intelligence, fertility and developmental milestones are normal in OCA, but poor vision and photophobia limit their educational attainment so that they are often restricted to unsuitable outdoor jobs that compound their problems. Together with the harsh African sun, the cruel myths about the aetiology of albinism, skin cancer and poor vision severely compromise the quality of life of the African OCA [7-9].

Current management options for the disabling visual defects of OCA consist of early detection and correction of refractive errors, use of sunglasses or special filter glasses for photo-aversion, and prismatic spectacle correction may be needed for abnormal head posture [10,11]. Strabismus surgery is often unnecessary but may be performed to

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improve peripheral visual fusion fields. With the best of efforts, vision may be improved to 20/40(6/12) whilst a significant number remain at about 20/200(6/60) [1,3,4]. Low vision aids (LVA) are therefore integral to the management of the patient with OCA. These devices are necessary in order to optimise the visual function for activities of daily living.

Review of available literature has failed to elicit the prevalence of ocular structural abnormalities in OCA. We therefore sought to investigate structural anomaly in the eyes of our visually impaired OCA with severe sun damaged skin, using the B-Mode ultrasound [12].

B-Mode ultrasound scan of the eyes is now a frequently requested examination used to produce a two-dimensional, cross-sectional view of the eye and orbit. It is otherwise called brightness scan [13-15]. It is safe (does not involve the use of ionizing radiation), readily available, easy and rapid to perform, affordable, non-invasive and can be repeated often. Furthermore, it is well tolerated by patients. It is done using high frequency transducers ranging from 7.5 MHz-15 MHz as this will distinctly demonstrate the internal ocular anatomy being a superficial fluid-containing structure.

The eye with its fluid content and superficial location is ideal for ultrasonic examination as the fluid provides a perfect acoustic window producing images with excellent detail. Ultrasound is non-ionizing, cheap, readily available, reproducible and well tolerated by patients. The need for pupillary dilatation and direct ophthalmoscopy to examine the posterior segment of the eye is obviated by the use of ultrasound. It is the only practical method of obtaining images of the posterior segment of the eye when the light conducting media are opaque [14].

The normal eye appears as a circular hypoechoic structure. Its cornea is seen as a thin hypoechoic layer parallel to the eyelid. The anterior aqueous chamber is filled with anechoic fluid and is bordered by the cornea, iris, and the anterior reflection of the lens capsule. The iris and ciliary body are seen as echogenic linear structures extending from the peripheral globe towards the lens. The normal lens is anechoic. The normal posterior vitreous chamber is also filled with anechoic fluid in the young but may show few internal echoes in the elderly. The choroid and retina appear as a single thin echogenic line indistinguishable from each other outlining the back of the eye. The optic nerve is visible posteriorly as a hypoechoic linear band coursing through the hyperechoic retrobulbar fat radiating away from the optic globe [12,13].

Materials and Methods

All of the 57 visually impaired Nigerians with a diagnosis of oculocutaneous albinism (OCA), aged 15 to 62 years who currently attend the Dermatology Clinic of Lagos State University Teaching Hospital (LASUTH) Ikeja, Lagos, Nigeria, were referred for ocular scan at the Radiology Unit of the same hospital. Fifty seven randomly selected age and sex matched normo-glycaemic Nigerians without albinism were recruited into the study from other referrals for routine ultrasonography. Demographic data were recorded.

The OCA had been diagnosed with low vision at the Eye department of the same hospital. Their distance visual acuity (VA) was assessed with Snellen's Distance Visual Acuity Chart at a distance of 6 meters, and the near vision assessed with Snellen's Near VA chart. Refractive error was measured with an auto-refractometer and subjective refraction was performed using the trial lenses box. Ophthalmoscopy and assessment for low vision were also performed. World Health Organization (WHO) classification of visual acuity, visual impairment

and blindness was used [16]. Normal vision is categorized within 6/6 to 6/18, moderate visual impairment $\leq 6/18$ to 6/60, severe visual impairment $\leq 6/60$ to 3/60, blindness $\leq 3/60$ in the better eye.

Real time B-Mode ocular scans were conducted using a linear array multifrequency probe (5.5 MHz-10 MHz) of a dynamic imaging General Electric LOGIQ 5 EXPERT machine.

The procedure was explained to the subjects and parents to ensure cooperation while scanning. We used the contact method of examination, with the patient lying supine, the probe placed directly on the closed eyelid with copious amount of coupling gel. The eyes were examined initially in the neutral position, and then in both the transverse and longitudinal planes. Subsequently, all participants were asked to move the eyes in all directions of gaze to further assess the eyes. We examined the status of the sclera, ciliary body, lens, vitreous, choroid, retina, the optic nerve and the macula. The average scan time was ten to fifteen minutes each. On completion of scanning, excess gel was removed with a swab.

Data obtained was entered into a Microsoft Office Excel spreadsheet and statistically analyzed using Statistical Package for Social Science (SPSS, Chicago, IL, USA) Version 18. Frequency tables were generated for variables and analysis was done using simple descriptive statistics. Ethics approval for the study was obtained from the Ethics Committee of the hospital. All participants voluntarily signed the informed consent form while parents signed for their children who were under 18years. In addition, assent for the procedure was obtained from children aged 8 years to 18 years. Signed consent was obtained for photography.

Results

All OCA were healthy normo-glycaemic, without any comorbidity and their average Body Mass Index (BMI) was 25. The controls were similarly matched. There were 30 (52.6%) males and 27 (43.4%) females. Mean age was 24.6 years. Majority (55/57) were less than forty years of age. Seven families had more than one OCA per family. All controls were normally pigmented without excessive cutaneous solar damage.

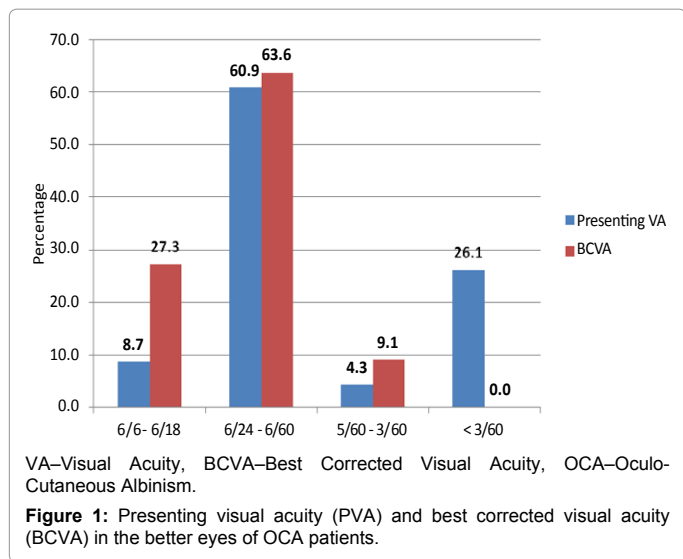
Skin examination of the OCA

All OCA admitted past history of several sun burn episodes. Solar Elastosis was universal in all 57 (100%) while 22/57 (38.5%) had at least one past skin cancer surgery and 33/57 (57.9%) had multiple solar keratoses.

Visual assessments

The presenting visual acuity in the better eye of the OCA patients is presented in figure 1. 8.7% had normal vision (6/6-6/18), majority (60.9%) had moderate visual impairment (6/24-6/60), 4.3% had severe visual impairment (5/60-3/60), while 26.1% were blind (presenting VA of $<3/60$). However, 68.2% of the patients had an improvement of at least one line of Snellen chart in their VA after refraction and/or low vision assessment. Proportion of those with normal vision increased to 27.3%, with no patient in the blindness category after refraction and assessment of best corrected visual acuity (BCVA).

Majority (52.3%) of the patients had myopic astigmatism, 18.2% had hypermetropic astigmatism, 13.6% had myopia, 11.4% hypermetropia, and 4.5% had simple astigmatism. In the myopic group, the minimum error was -1.00 diopters sphere (DS), maximum was -20.00 DS, with an average error of -7.50 DS. In the hypermetropic group, the minimum error was +0.50 DS, maximum was +7.25 DS, with an average of +2.5 DS. In the astigmatic group the range was -0.75 to -5.75 diopters



cylinder (DC), with an average of -3.00 DC. Strabismus was seen in 46 (80.7%) patients.

Most of the controls 48/57 (84.2%) had normal vision with presenting VA of 6/9, ranging from 6/5 to 6/18. There was no gender difference in the presenting VA of both groups.

Ocular scans

Among the OCA 53 (92.98%) had normal ocular scans, while 4(7.02%) had abnormal scans. The four abnormal scans which showed vitreous opacities were from 3 (75%) males and one female (Figure 2) (Table 1). Among the controls, fifty two (91.23%) had a normal ocular scans while 5 (8.77%) were abnormal. The five abnormal scans were four (7.02%) vitreous opacities (Figure 3) and one (1.75%) cataract (Figure 4) in a control patient and they were all males.

Inferential statistics did not reveal any significant difference in the data obtained from OCA and the controls.

Discussion

Our quest for vision enhancing correctable ocular structural abnormality in OCA was unproductive. Instead we found more structural abnormalities in the control population than in the OCA group. Particularly, we found no cataract among the OCA whereas one was found in the control group. The vitreous echoes which may be attributable to infective or inflammatory processes were more in the control group.

Ultraviolet radiation and light in the visible blue have been implicated as a cause of acquired cataract in the general population [15,16]. It is expected that the absence the protective melanin in the iris of OCA would predispose to cataract but our study has revealed a paradoxical absence of cataract in OCA.

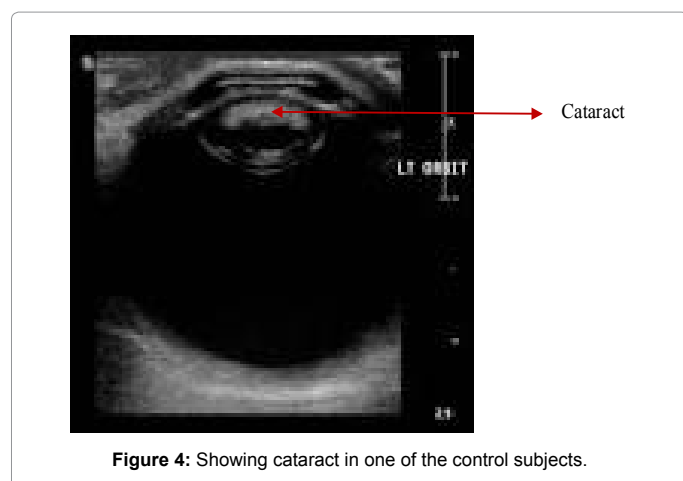
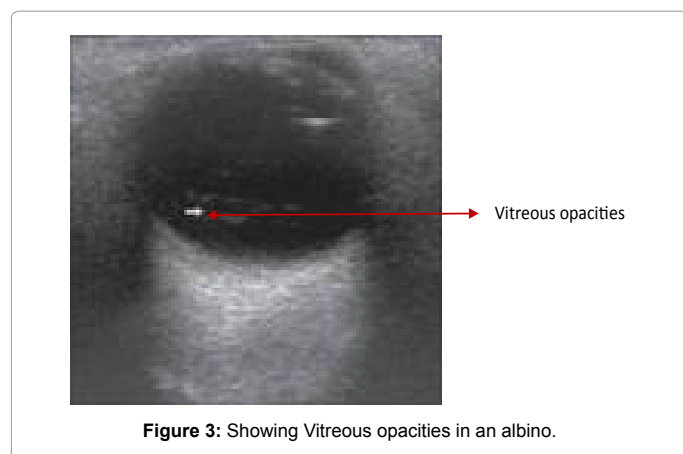
This may be because the OCA very early in life learns to use the eyelids as shield for the eyes or that the OCA does not live long enough to develop cataract. The latter is not supported by the fact that up to age 62 years our cohort of African OCA had no cataract. Again could it be that they kept all the rules of sun protection? This is not supported by evidence of considerable solar exposure manifested as extensive solar elastosis, previous excisions of basal cell carcinoma and squamous cell carcinoma from the face and neck of up to 30% of them.

We were inclined to consider the non-pigmentary functions of the melanocyte [17]. The melanocyte cell beyond production, transport and transfer of melanin also has other functions related to its interaction with other epidermal cells including the keratinocytes, the Langerhans cell, Merkel Cell and Fibroblasts. Although details of these interactions are still evolving, we do know that melanin augments

Variables	Albino		Control	
	Frequency	(%)	Frequency	(%)
Gender				
Male	30	52.6	30	52.6
Female	27	47.4	27	47.4
Total	57	100.0	57	100.0
Sex distribution of positive ocular scan				
Male	3	75.0	5	100.0
Female	1	25.0	0	0.0
Total	4	100.0	5	100.0
Age distribution of positive ocular scan	Albino		Control	
	Normal	Abnormal	Normal	Abnormal
<20	26	1	26	1
21-30	16	2	17	1
31-40	9	1	8	2
41-50	0	0	0	0
51-60	2	0	1	1
Total	53	4	52	5
Findings on ocular scan	Albino		Control	
	Frequency	(%)	Frequency	(%)
No Abnormality	53	92.98	52	91.23
Vitreous opacity	4	7.02	4	7.02
Cataracts	0	0.0	1	1.75
Total	57	100.0	57	100.0
Skin Examination				
Solar Elastosis	57	100	Nil	Nil
Lentigines	28	49.1	Nil	Nil
Solar Keratoses	30	52.6	Nil	Nil
Bowen's Disease	10	17.5	Nil	Nil
Basal Cell Carcinoma (BCC)	22	38.6	Nil	Nil
Squamous Cell Carcinoma (SCC)	15	26.3	Nil	Nil

Table 1: Descriptive analysis of ocular scan findings with skin examination in OCA and normally pigmented controls.





epithelial fibroblastic activity [17,18]. The high prevalence of keloids, an exaggerated fibroblastic response to skin injury, in the skin of color has been traced to the higher epidermal melanin [17,19]. Conversely the low prevalence of keloids in African OCA is considered to be related to absence of epidermal melanin [18,19].

One cause of ocular cataract is fibrous metaplasia of ocular lens epithelium [15]. Could it be that the absence of melanin also inhibits this fibrous metaplasia in the same manner that keloid formation is inhibited? Although this is statistically not significant in our study, we consider it clinically relevant to warrant further study.

Our study supports the current understanding that the functional visual impairment in albinism appears to have no correctable structural component [1].

The limitations of our study include the small available sample size of this rare condition, which robs this study of its statistical significance but perhaps not the clinical significance. Also, diagnosis of cataract was clinical. Slit-lamp examination, the conventional sensitive method of detecting cataract, was not done for the OCA group to ensure the same examination procedure for both control and test groups since this was a comparative study.

It is hoped that our study will stimulate more elaborate studies of the paradoxical absence of cataract in OCA.

Conclusion

Ultrasound scan reveals no correctable ocular structural abnormalities in OCA. The intriguing absence of cataract in African OCA is worthy of further investigation.

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