



Sociodemographic and visual characteristics of individuals with oculocutaneous albinism in Botswana



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Background: Understanding the characteristics of oculocutaneous albinism (OCA) in African populations is crucial for developing appropriate support strategies.

Aim: This study aims to comprehensively analyse the demographic and visual characteristics of individuals with OCA in Botswana, Africa.

Setting: This retrospective study was conducted at a private eye clinic in Botswana, Africa.

Methods: A retrospective data collection approach was employed to gather information from African patients diagnosed with OCA. Information on demographics, socio-economic status, education, occupation, medical history, category of visual impairment, visual characteristics such as contrast and glare sensitivity and central visual field integrity and low vision services received was extracted from medical records and analysed.

Results: The study involved 65 participants, primarily males (40, 61.5%), with a mean (s.d.) age of 29.2 (15.4) years. Most participants belonged to low (35, 53.8%) to middle-income (23, 35.4%) socio-economic groups. African-Tswana ethnicity was the most common among the participants (31, 47.7%). Nearly 80% had a best-corrected visual acuity (in better eye) worse than 6/12. Most participants had poor contrast sensitivity (47, 72.3%) and no defects in the central visual field (63, 96.9%). All participants experienced glare sensitivity (65, 100%).

Conclusion: The findings highlight the critical need for targeted interventions to address the prevalent visual impairments among individuals with OCA in Botswana, aiming to enhance their quality of life and socio-economic integration.

Contribution: These findings can inform interventions for OCA in Botswana, such as vision rehabilitation programmes, social support initiatives, and genetic counselling services.

Keywords: oculocutaneous albinism; demographic profile; clinical profile; Botswana; socio-economic status.

Introduction

Oculocutaneous albinism (OCA) is an inherited autosomal recessive disorder that affects skin, hair, and eye pigmentation because of mutations in the genes responsible for producing or distributing melanin.¹ People with OCA may have reduced visual acuity, nystagmus, strabismus, and photophobia, among other ocular conditions.^{2,3,4} Unfortunately, there is currently no known cure for OCA. Treatment is focused on managing the visual impairments (VIs) associated with the condition, which may include prescription spectacles or contact lenses, low vision aids, and specialised educational and vocational services.³

Varied prevalence of OCA has been reported across African countries, and different populations have been isolated. The mean prevalence reported was 1 in 4264 (1/1755 to 1/7900) African individuals.⁵ The most commonly reported condition was OCA2, an autosomal recessive condition observed in Southern Africa with a prevalence of 1 in 4000.⁵ Tswana village (isolate) from Botswana, with a population of 18000, reported a prevalence of 1 in 1300.⁶ Although OCA affects people worldwide, there is limited information on the visual function of individuals with OCA in Africa. This study aims to profile the sociodemographic and visual characteristics of people with OCA in Botswana, Africa.

Research methods and design

This descriptive study aimed to collect retrospective data from African patients who had visited Eyelight Optometrists, Tlokweng, Gaborone, Botswana, Africa. The selection of participants was

done through convenience sampling. The study included individuals with a confirmed diagnosis of OCA who had attended the clinic from the inception to May 2024, excluding those whose diagnosis was not OCA or had incomplete medical records. The study was approved by the Ministry of Health of the Republic of Botswana (HPDME 6/14/1). As the study was retrospective in nature, written consent was not obtained. However, patients at the private clinic provided consent during the registration process for using their unidentified data in scientific publications.

The data collection process involved reviewing the medical records of eligible participants to extract relevant information, such as demographics, socio-economic status, education level, occupation, medical history, category of VI,⁷ contrast and glare sensitivity, central visual field integrity, low vision services received, as well as best-corrected visual acuity (BCVA) for distance and near in the better eye. Distance visual acuity was assessed using a Snellen visual acuity chart (projection chart), while near visual acuity was evaluated using a continuous text near chart, with measurements taken at the patient's reading distance, which ranged from 20 cm to 40 cm, depending on individual preference. Contrast sensitivity was assessed using the Pelli-Robson chart, with results categorised as good (scores ≥ 2.0) and poor (scores < 2.0). Amsler grid was employed to assess central visual field integrity. Glare sensitivity was evaluated subjectively through detailed patient interviews, as a glare tester was unavailable. The collected data were then securely entered into a database for further analysis. The analysis used descriptive statistics to summarise the data using SPSS V20.

Ethical considerations

Ethical clearance to conduct this study was obtained from the Republic of Botswana Ministry of Health, Health Research Development Division (reference no.: HPDME 6/14/1).

Results

This retrospective analysis included 65 participants with a mean (s.d.) age of 29.20 (15.4) years and age range of 2–73 years. Most participants had BCVA in the better eye ranging from 6/12 to 6/60, with nearly 80% having worse than 6/12. Near vision analysis showed the majority of participants had near visual acuity of N6. Sociodemographic and visual characteristics of the participants are presented in Table 1 and Table 2, respectively.

Discussion

The retrospective analysis of 65 OCA participants offers significant insights into the study population's visual functions and sociodemographic characteristics. The study included a diverse participant pool, ensuring a broad representation of ethnic backgrounds. Males constituted a slightly higher proportion of the sample (61.5%), which is consistent with global trends observed in the prevalence of specific VIs.⁸ This disparity might be partially explained by

TABLE 1: Sociodemographic characteristics of the participants ($N = 65$).

Demographic characteristics	<i>n</i>	%
Sex		
Male	40	61.5
Female	25	38.5
Ethnicity		
African-Tswana	31	47.7
African-Bahrutshe	15	23.1
African-Baherero	2	3.1
African-Bakalagadi	2	3.1
African-Bakalanga	4	6.2
African-Banoka	2	3.1
African-Baserwa	1	1.5
African-Basherero	3	4.6
African-Basrwa	1	1.5
African-Batswapong	1	1.5
African-Bayei	3	4.6
Socio-economic status[†]		
Low	35	53.8
Middle	23	35.4
High	7	10.8
Educational level		
Basic	49	75.4
Degree	16	24.6
Occupation		
Student	22	33.8
Unemployed	20	30.8
Other (employed, self-employed, child)	23	35.4
Presence of other medical conditions		
Good health	62	95.4
Diabetes mellitus	2	1.5
Hypertension	1	3.1
Family history of albinism		
No	62	95.4
Yes	3	4.6

[†]. Socio-economic status: Low: Primary education or less, unemployed or sporadically employed, income below BWP 2000 or USD 200. Middle: Secondary education, small-scale business or stable low- to mid-level jobs, income BWP 10 000 or USD 1000. High: Tertiary education, consistent employment in mid-to-high-level positions or substantial business ownership, income above BWP 20 000 or USD 2000.

limited or unequal access to healthcare services, as men are often prioritised or more likely to seek medical attention conditions, potentially leading to their higher representation in the study.⁹ In addition, cultural and societal factors may influence the likelihood of men and women seeking diagnosis and treatment for VIs. However, further research is needed to understand the underlying factors contributing to this difference fully and to address potential biases in healthcare access and treatment. By investigating the specific reasons for the higher proportion of male participants, future studies can help inform strategies to improve healthcare equity for individuals with VIs, regardless of sex.

The ethnic diversity of the participants, primarily from different African communities, highlights the importance of considering cultural and genetic factors when examining visual health outcomes. This diversity provides a unique opportunity to explore how cultural perspectives and practices influence the examination and management of VIs. For instance, cultural beliefs and access to healthcare services can significantly influence an individual's decision to seek

TABLE 2: Visual characteristics of the participants ($N = 65$).

Visual characteristics	<i>n</i>	%
Category of visual impairment (VI)		
No VI ($\geq 6/12$)	5	7.7
Mild VI ($< 6/12$ but $\geq 6/18$)	19	29.2
Moderate VI ($< 6/18$ but $\geq 6/60$)	25	38.5
Severe VI ($< 6/60$ but $\geq 3/60$)	16	24.6
Contrast sensitivity (Pelli-Robson Chart)		
Good (Score: ≥ 2.0)	18	27.7
Poor (Score: < 2.0)	47	72.3
Glare sensitivity (Photophobia)		
Present	65	100.0
Central visual field (Amsler test)		
No defect	63	96.9
Defect present	2	3.1
Low vision services received		
Not required	48	73.8
Not willing	17	26.2

Note: Categories of visual impairment are according to ICD-11 for mortality and morbidity statistics [homepage on the Internet]. [cited 2024 Sept 30]. Available from: <https://icd.who.int/browse/2024-01/mms/en#1103667651>

medical help and adhere to treatment plans.¹⁰ Furthermore, genetic predispositions common within certain ethnic groups can offer insights into the prevalence and manifestation of specific visual conditions, guiding more tailored and effective screening and intervention strategies. Therefore, understanding these cultural and genetic nuances is essential in developing comprehensive approaches that enhance the health and well-being of diverse populations.

Most of the participants had a low to middle socio-economic status and basic education level, which reflects the broader socio-economic challenges faced by this population. In addition, a lack of awareness about albinism, as well as discrimination and stigmatisation against individuals with the condition could be contributed. These factors limit access to education and economic opportunities and hinder social integration and access to appropriate healthcare. Stigmatisation, in particular, can lead to social isolation and exacerbate mental health issues, necessitating a comprehensive approach that addresses both the medical and social challenges faced by individuals with albinism. These factors are critical determinants of health access and outcomes, including eye health.^{11,12}

A significant portion of the cohort did not require (48 out of 65) or were unwilling (17 out of 65) to receive low vision services. This discrepancy warrants further exploration to better understand and address the population's needs. Awareness and perception of VIs may play a crucial role, with some participants underestimating the severity of their condition or adapting to reduced acuity over time, leading to a perceived lack of need for services. Moreover, while all participants had access to low vision care, factors such as accessibility, availability, or cost may still have influenced their decision not to seek services, despite a genuine need.

Majority of participants had moderate VI, which is in line with previous publication from a cohort of Asian patients with OCA.^{13,14} This high prevalence of reduced visual acuity

highlights the need for targeted interventions to manage VIs in this cohort. It emphasises the impact of visual challenges on the daily lives of these individuals and how these impairments can significantly affect their quality of life, limiting their ability to perform everyday tasks and engage fully in social and economic activities.^{15,16}

The majority of participants in this study exhibited poor contrast sensitivity, aligning with the findings of previous research.¹⁷ This study's limitation lies in the absence of objective methods for evaluating glare sensitivity or photophobia. However, earlier research has shown that nearly 80% of individuals with OCA experience severe photosensitivity, which is consistent with the results observed in this study.³ For individuals with OCA, the intense discomfort from bright lights and the struggle to differentiate between contrasts can severely hinder their capacity to navigate surroundings, pursue educational and professional opportunities, and engage in community life. This highlights the need for a comprehensive approach to VIs, advocating for personalised interventions and support systems that address these specific challenges. It highlights the significance of inclusive strategies that improve mobility, accessibility, and social integration, empowering individuals with VIs to lead more enriching lives. The Amsler Grid test,¹⁸ a commonly used but less sensitive screening tool for identifying abnormalities in the central visual field, was used to assess the central visual field in this cohort and indicated that most participants had nearly normal central visual fields. However, static perimetry could have provided a more detailed assessment of the depth and extent of central visual field loss.

Healthcare professionals and policymakers should consider the findings when developing and executing strategies to prevent, identify, and manage VIs. Customised approaches that cater to the unique needs and circumstances of diverse populations can improve the effectiveness of such strategies, ultimately enhancing visual health outcomes and the overall quality of life for affected individuals.^{12,13,14}

The study's findings emphasise the importance of increasing awareness and education on visual health, especially in communities with limited access to healthcare services. Implementing community-based interventions and improving healthcare infrastructure can significantly reduce the impact of VIs. Further research should focus on exploring these interventions' effectiveness and outcomes.

Several limitations need to be taken into account when interpreting the results. Firstly, the study is retrospective, so it is impossible to establish a cause-and-effect relationship between demographic or clinical characteristics and visual acuity outcomes. This is because retrospective studies rely on existing records, which may not contain all the relevant data or be inaccurate, resulting in potential bias.

Secondly, the sample size of 65 participants, although adequate for preliminary observations, may not be large enough to generalise the findings across broader populations.

The broad age range and the variety of ethnic groups mainly represent the diversity within the sample. However, this also complicates the ability to draw specific conclusions about any demographic segment. Larger, more focused studies would be required to validate these findings and explore nuances within specific subgroups.

Lastly, while socio-economic status was considered, more detailed information about participants' access to healthcare, dietary habits, occupational exposures, or lifestyle choices could have offered more profound insights into the determinants of visual health. A more detailed breakdown or consideration of literacy levels could provide a richer understanding of the relationship between education and visual health.

Conclusion

There is a high rate of reduced visual function among the study's participants, highlighting the need for targeted interventions. Healthcare professionals and policymakers must consider the diverse socio-economic and ethnic backgrounds of individuals when designing health initiatives. Awareness and education on the condition (OCA) itself is also important in addressing the discrimination and stigma associated with the condition as this also influences whether affected individuals seek assistance and are compliant with management strategies. Future research is required to explore and mitigate the impact of VIs through community-based interventions and enhanced healthcare services.

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Competing interests

The authors declare that they have no financial or personal relationships that may have inappropriately influenced them in writing this article.

Authors' contributions

G.M. contributed to the design and planning of the study, analysing, writing the original draft, and reviewing the article. T.P. was involved in the design and planning of the study, data collection, analysis, and reviewing the article.

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Data availability

The data that support the findings of this study are available, upon reasonable request from the corresponding author, T.P.

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