Primary cutaneous malignancies in black African patients with oculocutaneous albinism in KwaZulu-Natal Province, South Africa

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Background. Individuals with oculocutaneous albinism (OCA) face a significantly heightened risk of developing skin cancer owing to increased sensitivity to ultraviolet (UV) radiation. Data on the epidemiology and geographical distribution of cutaneous malignancies among persons with albinism (PWA) in KwaZulu-Natal Province (KZN), South Africa, remain limited.
Objective. To investigate the demographic and regional patterns of primary cutaneous malignancies within this high-risk population.
Methods. We conducted a retrospective descriptive analysis of all PWA diagnosed with histologically confirmed primary cutaneous malignancies treated at a tertiary healthcare centre in KZN between January 2002 and June 2022. Sociodemographic, geographical and tumour characteristics were extracted from electronic medical records.
Results. A total of 221 patients (56.6% female, 43.4% male) with a mean (standard deviation) age of 37.9 (12.5) years contributed 618 histologically confirmed malignant lesions. 132/221 patients (59.7%) presented before 40 years of age. Female patients presented slightly earlier (37.1 (12.9 years)) than males (39.0 (12.0 years)). Geographical clustering was observed in eThekwini (43.0%), uMkhanyakude (20.8%) and King Cetshwayo (11.8%) municipalities. Squamous cell carcinoma (SCC) (50.2%) and basal cell carcinoma (BCC) (49.7%) were nearly equally distributed across all the districts; one cutaneous melanoma was identified.
Conclusions. In KZN, PWA tended to develop skin cancer at a younger age than the general population. Notably, the proportions of SCC and BCC were roughly equivalent – a finding that contrasts with historical data, which show a predominance of SCC. The high incidence of skin cancer in coastal and rural districts aligns with elevated ambient UV exposure, and highlights disparities in healthcare access within these regions. These findings highlight the urgent need for targeted screening programmes, improved photoprotection strategies and culturally sensitive education initiatives to reduce morbidity and mortality among this vulnerable population.

Keywords: oculocutaneous albinism, skin cancer, KwaZulu Natal

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Oculocutaneous albinism (OCA) is a heterogeneous group of autosomal recessive genetic disorders characterised by a partial or complete absence of melanin in the skin, hair and eyes.[1] Although skin cancers are rare among black Africans,[2-4] the absence of melanin's protective function in persons with albinism (PWA) significantly increases their vulnerability to the harmful effects of ultraviolet (UV) radiation, thereby elevating their risk of developing cutaneous malignancies.[4] Challenges faced by individuals living with OCA in KwaZulu-Natal (KZN), a province situated on the east coast of South Africa (SA), are not limited to the skin. African cultural beliefs and stigma,[2-6] unfavourable climate conditions[7-9] and associated health risks[10-12] with limited access to specialised healthcare[13,14] all contribute to the vulnerability of these individuals in the province.

Primary cutaneous malignancies are a major cause of mortality in young Africans with OCA. They are diagnosed at an earlier age, with more advanced stage of disease on first presentation and a higher rate of recurrence when compared to the general population without OCA.[15-17] The incidence of squamous cell carcinoma (SCC) in the general black African population varies greatly between regions,[18] with this risk increased 1 000-fold in PWA.[19,20] In KZN, these individuals live in regions where they are exposed to high levels of UV radiation[7,9] and therefore are more prone to developing skin cancer due to the absence of melanin pigmentation.[21]

Premalignant lesions such as actinic keratoses are frequently observed in PWA, and represent important precursors to invasive SCC.[17,22] A high prevalence of actinic damage in this population has been documented,[11,23] and highlights the burden of cumulative UV radiation exposure and the need for earlier detection and management.

To better understand the epidemiology and the relationship between geographical location and the incidence of skin cancers among PWA in KZN, we conducted a retrospective analysis of 221 PWA who were treated for a total of 618 cutaneous malignancies at our facility. This information will identify high-risk regions of the province where more intensive screening and referral programmes are required. Poor patient awareness and limited access to specialised healthcare services continue to be a problem for PWA living in rural communities in the province.[14]

Methods

Study design

A retrospective descriptive study was conducted involving patients diagnosed with OCA who received treatment at a tertiary healthcare centre for primary cutaneous malignancies confirmed by histopathology.

Study area and population

The study was conducted in KZN, a province situated on the eastern coast of South Africa (–28.53°N 30.89°E). KZN has a population of ~11 065 240, distributed across 54 municipalities, including 10 district municipalities and 1 metropolitan municipality (eThekwini).

The coastal regions of the province experience a subtropical climate characterised by high ambient UV radiation throughout most of the year. The average annual maximum temperature for eThekwini (29.87°S 31.06°E;), the province's only metropolitan municipality, is 25.8°C.

Inclusion and exclusion criteria

Patients included in the study were those with a confirmed diagnosis of OCA and histologically verified primary cutaneous malignancies who presented to the healthcare facility between January 2002 and June 2022. Patients lacking complete clinical or histopathological records were excluded.

Data collection

Sociodemographic data were collected retrospectively from the electronic medical records of eligible patients. Histopathology reports were reviewed to extract information regarding the type and anatomical site of each cutaneous malignancy.

Statistical analysis

Collected data were entered and encoded using Excel (Microsoft, USA). Descriptive statistics were used to summarise demographic and clinical data. Mean values with standard deviations (SDs) and 95% confidence intervals (CIs) were calculated for continuous variables such as age and lesion counts. Categorical variables (sex and lesion type) were summarised as frequencies and percentages.

Ethical considerations

Ethical approval was obtained from both the University of KwaZulu-Natal and provincial ethics committees (ref. nos BREC/00004696/2022 and KZ202210008).

Results

Demographic details

A total of 221 patients were identified and included in the study. Appendix Table S1 shows the sex, mean age and age ranges distributed within the districts of KZN. There were 125 females (56.6%) and 96 males (43.4%), yielding a female-to-male ratio of 1.3:1. The mean (SD) age at first presentation was 37.91 (12.5) (range 13 - 80) years, with the majority (55.2%) diagnosed between the ages of 20 and 39 years; 59.7% of patients presented before the age of 40 years. The age distribution by gender is illustrated in Fig. 1. Female patients presented marginally earlier than male patients (37.1 years v. 39.0 years). The mean age of presentation for patients with SCC was 37.4 years, and 38.4 years in those patients with BCC.

Epidemiology

Geographically, most patients (n=95, 43.0%) originated from eThekwini municipality, followed by uMkhanyakude (n=46, 20.8%) and King Cetshwayo (n=26, 11.8%) districts (Fig. 2). Smaller patient contributions came from iLembe (n=18), Zululand (n=16), Ugu (n=14) and Harry Gwala (n=3), and single cases in uThukela and uMzinyathi. UMkhanyakude had the highest clinic-based prevalence (6.67/100 000) and was the only district noted to have a male predominance of patients. EThekwini municipality was the largest contributor of patients to the cohort, yet represented one of the lowest clinic-based prevalence rates (2.24/100 000), indicating a relatively small number of patients despite the metropolitan's large population. Fig. 3 shows the clinic-based prevalence rates for the six most represented municipalities.

A total of 307 BCCs (49.7%) and 310 SCCs (50.2%) were identified, producing an overall BCC:SCC ratio of 1:1. Fig. 4 illustrates the BCC:SCC lesion ratios and mean number of lesions per patient across the six most represented districts in the study. Zululand had the lowest lesion ratio (0.37; 95% CI 0.18 - 0.82), with the highest ratio noted in iLembe (1.88; 95% CI 0.85 - 4.25). The remaining districts clustered around parity (1.0) with moderately narrow CIs, which suggests an approximately equal frequency of lesions per patient. The highest lesion burden was seen in Ugu district (3.25 lesions per patient). One case of cutaneous melanoma (CM) (0.2%) was identified in the right lower limb of a 26-year-old female in uMkhanyakude district. SCC incidence was evenly distributed across age groups, whereas BCC predominated in patients ≥40 years of age.

Discussion

This study represents the largest single-centre study of African PWA and primary cutaneous malignancies. Currently, no prevalence data for OCA in KZN exists, and in SA in general, epidemiological data remain sparse and geographically limited. Localised studies highlighting significant regional variation of the condition with prevalence rates vary from 1 in 2 239 among the VhaVenda population,[24] 1 in 3 900 in Soweto, Johannesburg[25] and 1 in 6 000 in the Nongoma District of KZN.[26] The estimated prevalence of OCA in sub-Saharan Africa ranges from 1 in 4 000 to 1 in 7 000.[26-28]

The age of presentation is critical in understanding the burden of skin cancer in PWA, as earlier onset often reflects higher vulnerability and exposure. The mean (SD) age at first presentation in our study (37.9 (12.5) years) and the predominance of patients in the age range of 20 - 39 years (55.2%) highlights the high burden of skin cancer at a younger age in this vulnerable population. This aligns with reports from other African studies – similar mean ages were noted in studies conducted in Nigeria[29,30] and Togo,[31] where skin cancer typically presents between the third and fourth decades of life. The youngest mean age (26.5 years) was reported in the Democratic Republic of Congo,[32] further highlighting that there is a consistently earlier onset of non-melanoma skin cancer in African PWA as opposed to non-albino populations. A cohort from the Free State Province in SA demonstrated a higher mean age at skin cancer diagnosis of 48 years,[23] which may reflect regional differences in access to healthcare, occupational exposures or health-seeking behaviour. Zululand district presented with the lowest mean (SD) age of 30.9 (9.12) years, younger than the overall cohort average (37.9 (12.5) years). It was previously reported that African PWA predominantly developed skin cancers before the age of 30, and seldom survived beyond this age.[33] A report by the United Nations claimed that skin cancer due to sun exposure caused death in 98% of PWA before the age of 40 years.[34] This supports the importance of timely access to care for young individuals who are at risk of presenting with advanced disease owing to a delay in diagnosis.

The female predominance in our study (56.6%) reflects a trend noted in Brazil[35] and several African countries.[23,29,36-39] These variations likely reflect local gender roles, with rural women disproportionately affected owing to social and cultural practices involving outdoor labour and informal farming. Interestingly, female patients also presented with skin cancer earlier than males, with this pattern consistent across most districts of KZN. This finding questions the belief that females face greater barriers to early healthcare access, in part due to their economic dependence, societal norms and household and caregiving responsibilities.[40] Females

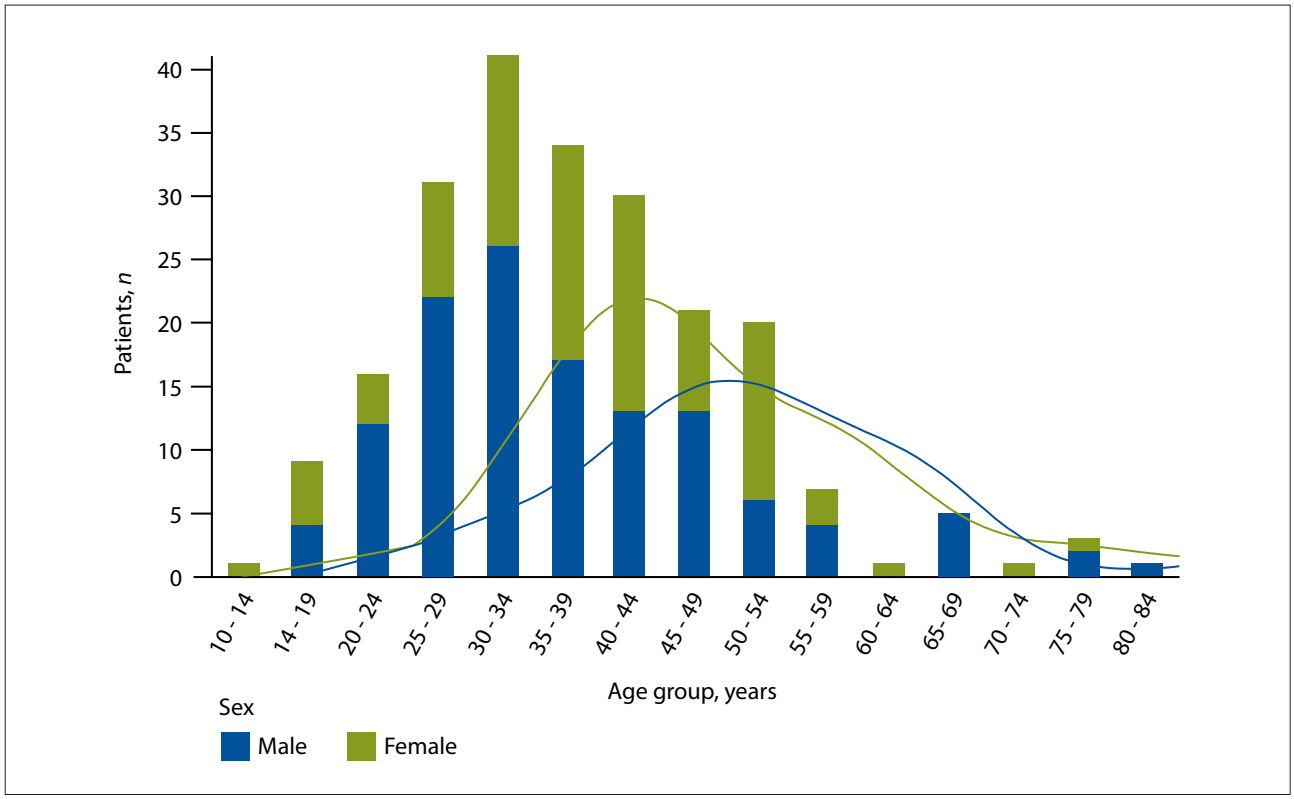

Fig. 1. Distribution of patients by age and sex.

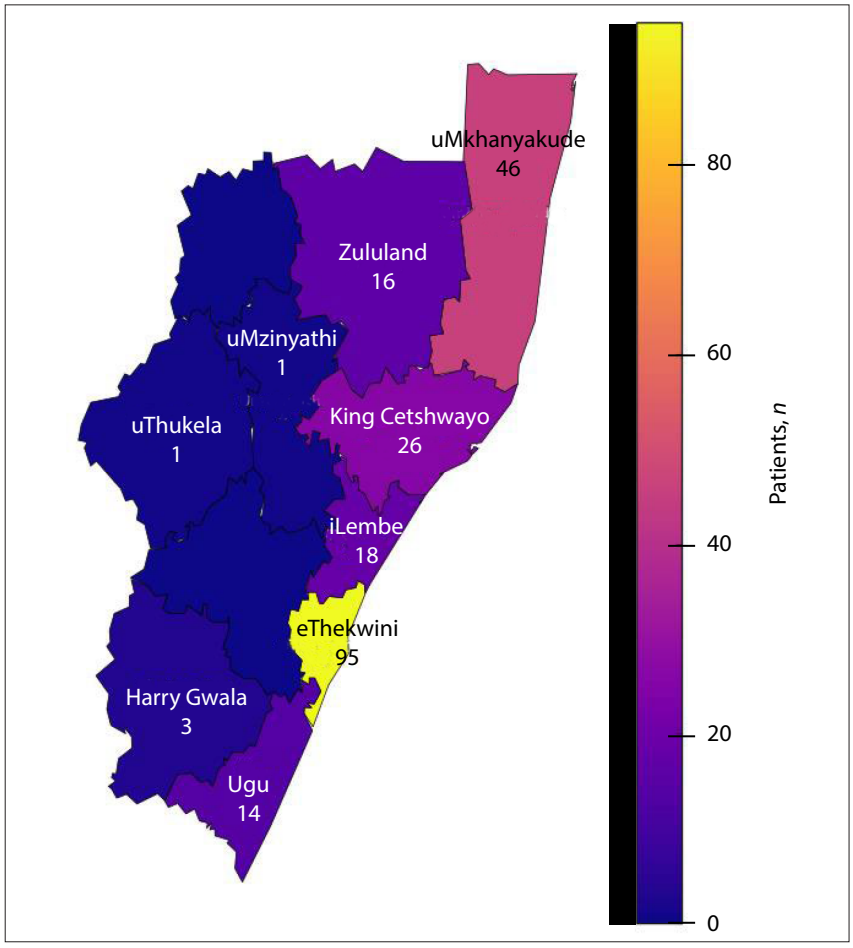

Fig. 2. Patient distribution by district in KwaZulu Natal Province, South Africa.

may in fact demonstrate greater health-seeking behaviour than males, a pattern consistently reported in both international[41] and local SA studies.[14,23] Despite this, there are significant intersectional challenges faced by female PWA living in KZN – a province where cultural factors also play a critical role. Stigmatisation and harmful myths surrounding albinism may contribute to social isolation, impairing an individual's willingness to seek healthcare regularly.[6,40]

In KZN, geographical clustering and gender differences in skin cancers among PWA reflect a complex interplay between environmental, occupational, cultural and socioeconomic factors, which are all compounded by disparities in healthcare accessibility. Although high UV radiation in the coastal regions contributes significantly to the risk of skin cancer, occupational factors are particularly relevant. Many residents in rural communities are engaged in outdoor activities and informal labour without adequate photoprotection, with limited access to shade infrastructure, thereby increasing their exposure to cumulative UV damage and malignancy risk.[42]

One patient originated from Alfred Nzo district in the Eastern Cape Province. This single case reflects a pattern of referral across provincial boundaries for specialist care, highlighting the need for regional collaboration in managing PWA.

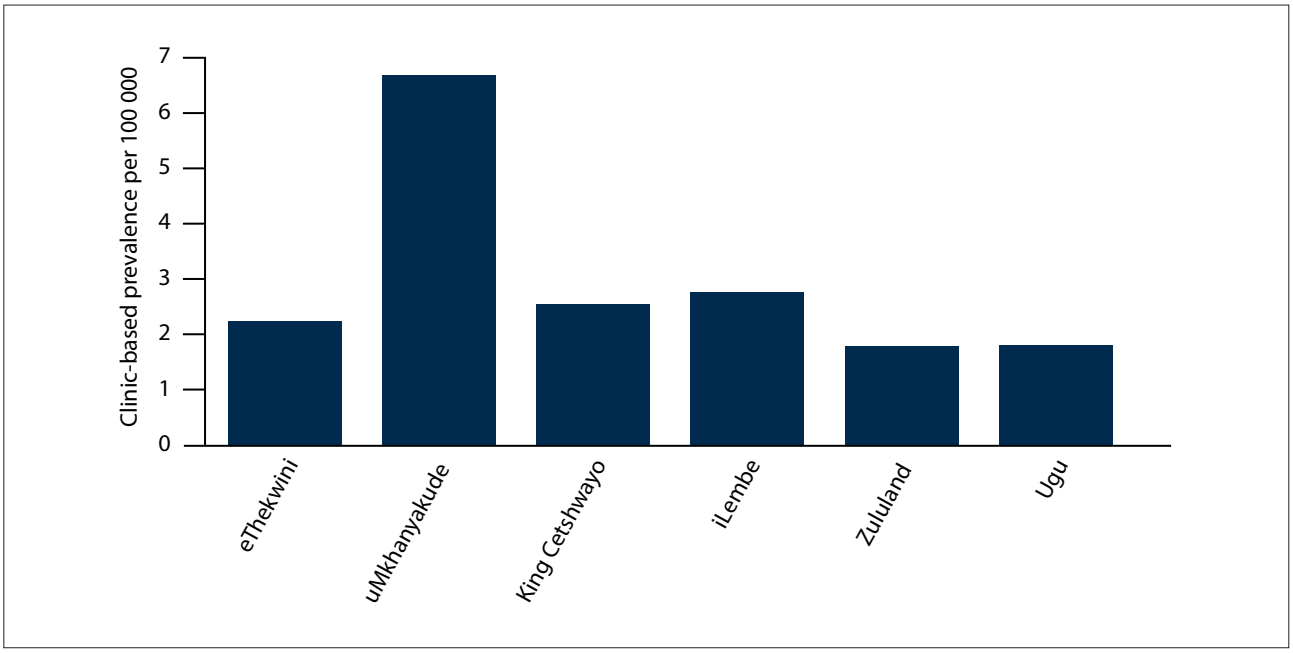

Fig. 3. Clinic-based prevalence of oculocutaneous albinism by district in KwaZulu-Natal Province, South Africa.

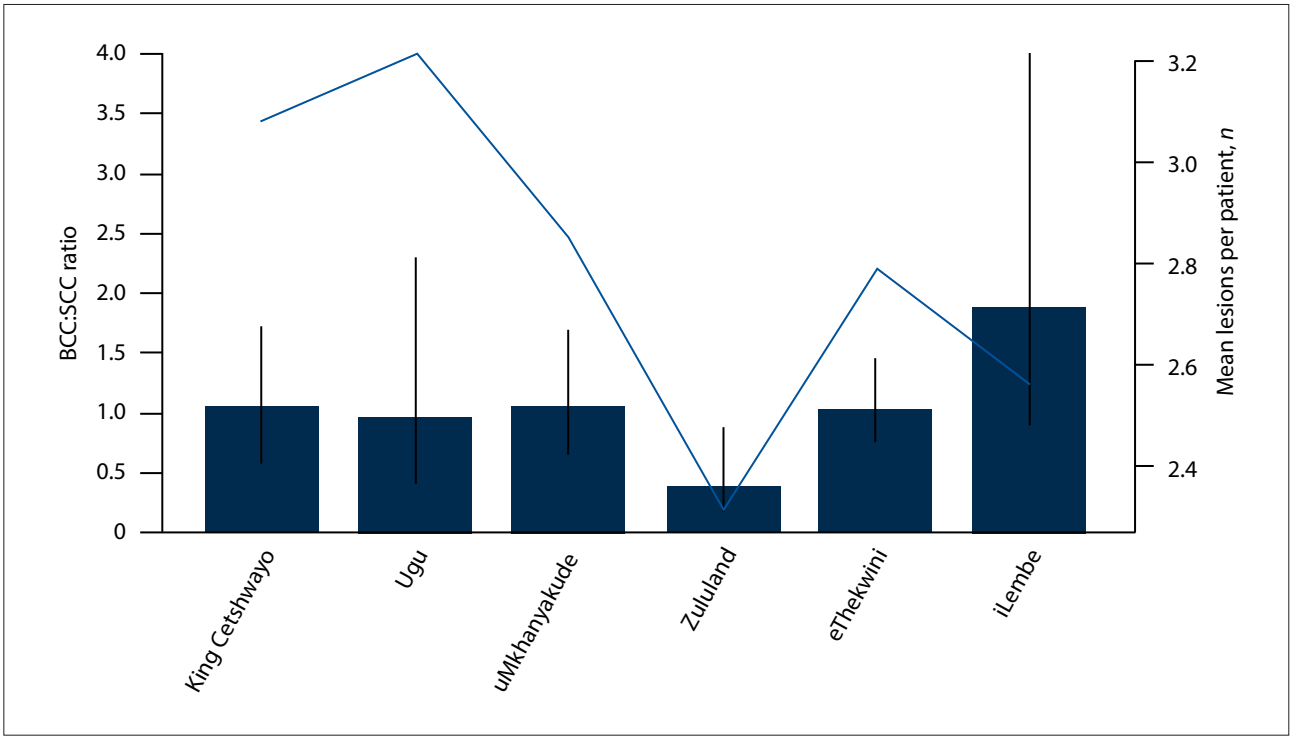

Fig. 4. Ratio of basal cell carcinoma (BCC):squamous cell carcinoma (SCC) and mean number of lesions per patient by district in KwaZulu-Natal Province, South Africa (95% confidence interval).

Healthcare accessibility has a significant influence on the morbidity of PWA with cutaneous malignancies. This is particularly evident in KZN, where substantial disparities exist between rural and urban municipalities. Patients referred from rural coastal municipalities were the most represented in this study (54.3%), compared with 43.0% referred from the more urban eThekwini metropolitan area. Urban areas generally offer better healthcare infrastructure with easier accessibility,[43] providing patients in these regions with more regular dermatological screening, contributing to earlier diagnosis and management of skin cancers. Conversely, rural districts in the province are faced with greater healthcare barriers, including fewer healthcare facilities, limited options for transportation and longer distances to travel for specialised care.[43] These districts, however, exhibited similar lesion counts per patient, in both SCC and BCC, with lower mean ages (37.0 years v. 39.3 years) at presentation compared with the urban metropolitan eThekwini area. This trend suggests that patients in these rural areas may be presenting earlier due to higher cumulative UV exposure or occupational risk factors.

Patterns of skin cancer

The coastal and north-eastern regions of KZN displayed a higher incidence of non-melanoma skin cancer. This geographical distribution aligns with a higher UV radiation exposure pattern, with the subtropical climate of these coastal regions and lower altitudes producing year-round UV radiation that is significantly higher than in inland regions.[7,9]

The nearly equal distribution of BCC and SCC observed among PWA in this study (1:1) represents a novel finding and a notable shift from historical data. Studies from sub-Saharan Africa have previously shown a SCC predominance,[12,16,20] with a similar SCC predominance in PWA demonstrated in historic SA studies.[17,19,27] Recent studies have shown either a more equal proportion of SCC and BCC,[22,23] or a more BCC-dominant distribution among PWA.[30,31,39,44] This suggests an epidemiological shift linked to improvements in healthcare access, with earlier detection through screening programmes that are allowing a greater number of BCC lesions to manifest as patients are living longer. These observations show that as healthcare improves, the patterns of skin cancer in PWA in Africa are beginning to parallel those seen in lighter-skinned populations where BCC typically predominates.[30] The lesion-type ratios in this study show heterogeneity between districts (0.37 - 1.88). However, there were no clear patterns or statistical associations identified.

The single case of CM in a young female represents a very small percentage of all histologically confirmed skin malignancies (0.2%). This is consistent with previous studies across sub-Saharan Africa, where CM in PWA is rarely reported. A systematic review identified only six cases of CM in 556 PWA.[45] Melanomas in the general black African population are most commonly of the acral lentiginous subtype, occurring most frequently in the non-sun exposed lower limb.[36,46] While UV radiation has been reported as a risk factor for CM in fair-skinned individuals, there is no recognised risk in darker-skinned people.[45] An analysis of the National Cancer Registry demonstrated a very low melanoma incidence among black Africans in SA, with no cases recorded in PWA.[47] It also reported that white people have >20 times higher incidence of CM than pigmented black Africans.[47] However, there appears to be no risk of CM as a result of the fair skin of albinism. The low incidence of CM in black PWA compared with whites and pigmented black Africans highlights an inconsistency that needs further investigation.

Prevention of skin cancers in PWA requires a comprehensive public healthcare approach. Interventions directed at behavioural modifications, such as the avoidance of midday sun exposure, physical protection in the form of protective clothing, hats and shade infrastructure, and the use of sunscreen from an early age, are all essential in reducing UV-induced skin damage. These measures need to be integrated into community education initiatives and tailored to the cultural contexts in KZN.

Study limitations

This study is limited by its retrospective design and reliance on tertiary level hospital records. This introduces potential selection and referral bias, as patients with advanced disease are more likely to be referred. The true prevalence of cutaneous malignancies among PWA in KZN may be underestimated, particularly in districts with limited access to referral pathways. Community-based prevalence studies are therefore needed to address these gaps.

Conclusion

This study highlights a significant burden of cutaneous malignancies among PWA living in KZN. There are geographical differences in terms of gender, age at first presentation and pattern of skin cancer. The high prevalence of OCA in select SA communities, combined with the elevated risk of UV-induced skin cancer, suggests an urgent need for comprehensive public health strategies. These should include targeted community and gender-specific education, improved access to dermatological care and the implementation of formal screening programmes – particularly in high-risk rural and coastal districts – to reduce morbidity and early mortality in this vulnerable population.

Data availability. The data used for this study are available from the authors upon reasonable request.

Declaration. This study project was led by PB and completed in fulfilment for his MMed (Plastic and Reconstructive Surgery) degree at the University of KwaZulu-Natal, under the supervision of AM.

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Author contributions. PB and AM designed and conceptualised the idea for the study. Data collection and manuscript drafting was done by PB, with revision of the manuscript for clinical validity done by AM. Both authors read and approved the final version of the manuscript.

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Conflicts of interest. None.

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