

## Communication

# Oculocutaneous Albinism in Northern Madagascar: Clinical Burden, Social Stigma, and Impact of a Community-Based Photoprotection Program

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## Abstract

Oculocutaneous albinism (OCA) increases susceptibility to ultraviolet (UV) skin damage, skin cancer risk, and psychosocial burden. Data from Madagascar are lacking. We conducted a six-month pilot study (July–December 2024) in northern Madagascar (DI-ANA and SAVA regions). Forty-one individuals with OCA were enrolled. Baseline socio-demographic, clinical, and behavioral data were collected through interviews and dermatological examinations. A structured program provided education, culturally adapted materials, and photoprotective resources, with monthly follow-up visits. The cohort included 22 males and 19 females, with a mean age of 18 years (range: 1 month–35 years). Actinic keratoses were present in 61% of participants, and invasive skin cancer in 4.9%. All patients had photophobia and nystagmus. Social discrimination was reported by 65.9%, with 12.2% describing severe abuse. Baseline photoprotection was inadequate: 43.9% reported no protective practices, 7.3% used sunscreen, and 19.5% avoided midday sun. Follow-up was completed by 20/41 patients (48.8%). Among completers, paired analysis showed a decrease in sunburn prevalence from 95.0% to 10.0% ( $p < 0.0001$ ), an increase in regular sunscreen use from 0.0% to 100.0% ( $p < 0.0001$ ), use of protective clothing from 35.0% to 80.0% ( $p = 0.0039$ ), and adoption of behavioral strategies from 15.0% to 50.0% ( $p = 0.0156$ ). This first study on OCA in northern Madagascar demonstrates a high burden of UV-related dermatoses and stigma. A low-cost community intervention significantly improved photoprotection. Wider implementation could reduce morbidity and enhance quality of life in resource-limited settings.

**Keywords:** oculocutaneous albinism; Madagascar; skin cancer; photoprotection; stigma; pilot study



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## 1. Introduction

Oculocutaneous albinism (OCA) is a rare autosomal recessive disorder characterized by varying degrees of hypopigmentation of the skin, hair, and eyes, accompanied by ocular features such as nystagmus, misrouting at the optic chiasm, foveal hypoplasia, and reduced visual acuity [1]. To date, eight subtypes of OCA have been identified, each associated with specific mutations affecting the melanin biosynthesis pathway and exhibiting variable

clinical severity [1]. Although OCA occurs in all ethnic groups, it has a disproportionately greater medical and social impact among individuals of African descent [1].

Due to melanin deficiency, individuals with OCA are particularly susceptible to ultraviolet (UV) radiation. Actinic keratoses (AKs) frequently develop on sun-exposed areas, especially the head and neck, from early childhood as a result of cumulative UV exposure, leading to a markedly increased risk of skin cancer [2,3]. Studies have reported up to a 1000-fold higher risk of squamous cell carcinoma (SCC) compared to the general population [4–7]. Basal cell carcinoma (BCC) is the second most common malignancy [8], while cutaneous malignant melanoma (CMM) is rare but has been documented [8]. Occupational exposure (e.g., farming, trading, herding) and environmental factors such as proximity to the equator and high altitude further amplify the risk [8–10]. Overall, life expectancy in individuals with OCA is significantly reduced, primarily due to skin cancer developing between the second and fourth decades of life [11,12].

In Africa, the etiology of OCA is often misunderstood and intertwined with myths and superstitions, contributing to stigma, social exclusion, and human rights violations [1,13]. In Madagascar specifically, the prevalence of albinism remains unknown due to the absence of systematic data collection [14]. Genetic testing is rarely available, except in a few specialized centers. A clinical study from South Africa demonstrated that certain OCA subtypes more prevalent in African populations can often be distinguished based on clinical features alone, thanks to genotype–phenotype correlations that, while not entirely precise, are useful in resource-limited settings [15].

Finally, the burden of sun-induced dermatoses among individuals with OCA is poorly documented, particularly in rural areas.

In Madagascar, as in many other African countries, access to photoprotection resources is extremely limited and awareness of preventive strategies remains scarce, particularly among vulnerable populations such as individuals with albinism.

This observational study had three main objectives: (1) to collect clinical and demographic data on the OCA population in the DIANA and SAVA regions of northern Madagascar; (2) to evaluate their baseline knowledge, behaviors, and skin health status; and (3) to assess the impact of an intervention program incorporating education, clinical care, and the provision of sun protection resources.

## 2. Materials and Methods

The study was conducted over a six-month period, from July to December 2024, in the northern regions of Madagascar (DIANA and SAVA), within the framework of the *Alma Mater Ticinensis* “Progetto Professionalità 2023–2024” grant. The project was hosted at Polyclinique NEXT in Diego Suarez, a health facility supported by the Italian NGO NEXT. The study was purely observational and non-interventional, consisting of clinical examinations, structured interviews, and the delivery of preventive and educational measures that are fully consistent with good clinical practice. The study was conducted in accordance with the Declaration of Helsinki (1975, revised 2013).

The multidisciplinary team consisted of the lead author (RD, a dermatology resident), an Italian nurse, a Malagasy–French interpreter, and an educator living with albinism, who played a key role in patient communication and health education.

Participants were eligible if they had a clinical diagnosis of OCA, resided in either the DIANA or SAVA regions, and had sufficient access to our fixed or mobile services.

Potential participants were identified through community associations for persons with albinism, local health centers, and word of mouth. Eligible individuals were invited to participate during mobile outreach visits or at our fixed clinic.

At the first visit, each patient underwent a structured interview conducted in Malagasy with the assistance of a local translator. The interview was designed to collect sociodemographic data, assess awareness of albinism, evaluate understanding of sun-related risks, document previous sunburn history, and record experiences of social discrimination based on the patients' responses.

A comprehensive dermatological and dermoscopic examination followed, including assessment of phototype, presence of dendritic freckles, actinic keratoses, skin cancers, strabismus, photophobia, and visual acuity.

Immediately after the medical evaluation, patients received an individualized educational session explaining the genetic basis of albinism and the importance of sun protection. As many participants were not literate, the education was delivered orally and interactively; those able to read were also provided with a bilingual brochure (Malagasy and French).

Each patient received a complete photoprotection kit containing SPF 50+ sunscreen, a wide-brimmed hat, UV-filtering sunglasses, and long-sleeved clothing donated by supporters in Italy. Education at the initial visit was provided one-on-one; during follow-up visits, it was often delivered in small groups to foster discussion and peer support.

Community outreach activities and collaboration with local networks led to the identification of 49 patients with OCA. Of these, 41 met the eligibility criteria and were examined in person by the medical team: 17 at Polyclinique NEXT and 24 through a mobile clinic deployed in the SAVA region.

Of the 41 patients, 20 returned for monthly follow-up visits over the six-month period. These follow-up sessions included clinical skin checks, reinforcement of photoprotection education, and provision of additional sunscreen.

Clinical data were collected through direct observation and photographic documentation. Informed consent was obtained from all participants or legal guardians. For illiterate participants, consent was obtained orally in the presence of a witness and documented by the study team.

Dichotomous variables were summarized as  $n/N$  (%). Baseline summaries refer to all enrolled participants ( $N = 41$ ). Inferential paired comparisons were restricted to participants who completed the 6-month follow-up ( $n = 20$ ) and were conducted using the exact McNemar test on  $2 \times 2$  transition tables calculated within the same individuals, reporting discordant pairs and two-sided  $p$ -values

### 3. Results

The study included 41 participants with OCA from the DIANA and SAVA regions: 53.7% (22/41) were male and 46.3% (19/41) female. Sixteen (39%) were children (<14 years), 4 (9.8%) were adolescents (14–18 years), and 21 (51.2%) were adults (>18 years). The mean age was 18 years (range: 1 month–35 years). None of the participants had previously undergone dermatological or ophthalmological evaluation, nor had any received surgical treatment for skin lesion removal.

A family history of OCA was reported in 60.9% (25/41), suggesting some degree of awareness within family structures. However, 63.4% (26/41) had no understanding of OCA as a genetic condition, and among those claiming some awareness, none were able to explain its hereditary basis.

Dermatological examination revealed dendritic freckles in 12.2% (5/41) and actinic keratoses in 61.0% (25/41), rising to 87.5% (21/24) among those older than 20 years. Two cases of invasive skin cancer were diagnosed through histopathological examination (4.9%, 2/41). Additional clinical characteristics are summarized in Table 1: hair color (29.3% [12/41] straw yellow; 70.7% [29/41] yellow), eye color (80.5% [33/41] blue/green; 19.5% [8/41] brown), photophobia (100%), strabismus (24.4%, 10/41), and nystagmus (100%).

**Table 1.** Demographic, clinical phenotypic and social characteristics of patients ( $n = 41$ ) at the first visit. Values are presented as percentages with absolute numbers in parentheses.

|                                                          |                                                                                                                                                                  |
|----------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Gender</b>                                            | 53.7% (22/41) male<br>46.3% (19/41) female                                                                                                                       |
| <b>Age</b>                                               | Children (<14 years) 39% (16/41)<br>Adolescents (14–18 years) 9.8% (4/41)<br>Adults (>18 years) 51.2% (21/41)                                                    |
| <b>Family history of albinism</b>                        | 60.9% (25/41) positive<br>39.1% (16/41) negative                                                                                                                 |
| <b>Patient awareness of their condition</b>              | 63.4% (26/41) no<br>37% (15/41) partial                                                                                                                          |
| <b>History of sunburns</b>                               | 90.2% (37/41) yes<br>9.8% (4/41) no                                                                                                                              |
| <b>Presence of solar erythema at first visit</b>         | Severe 53.7% (22/41)<br>Mild 39% (16/41)<br>None 7.3% (3/41)                                                                                                     |
| <b>Sun protection strategies reported at first visit</b> | Wearing covering clothing 39% (16/41)<br>Avoiding midday sun exposure 19.5% (8/41)<br>Using sunscreen 7.3% (3/41)<br>No strategy 43.9% (18/41)                   |
| <b>Clothing at first visit</b>                           | Covering 22% (9/41)<br>Not covering 78% (32/41)                                                                                                                  |
| <b>Presence of dendritic freckles</b>                    | 12.2% (5/41)                                                                                                                                                     |
| <b>Presence of actinic keratoses</b>                     | 61% (25/41) among the total patients<br>87.5% (21/24) among patients over 20 years old                                                                           |
| <b>Presence of skin tumor</b>                            | 4.9% (2/41)                                                                                                                                                      |
| <b>Hair color</b>                                        | 29.3% (12/41) straw yellow<br>70.7% (29/41) yellow                                                                                                               |
| <b>Eye color</b>                                         | 80.5% (33/41) blue/green<br>19.5% (8/41) brown                                                                                                                   |
| <b>Presence of photophobia</b>                           | 100% (41/41)                                                                                                                                                     |
| <b>Presence of strabismus</b>                            | 24.4% (10/41)                                                                                                                                                    |
| <b>Presence of nystagmus</b>                             | 100% (41/41)                                                                                                                                                     |
| <b>Phenotype/genotype suspected OCA subtype</b>          | OCA2 with freckles 12.2% (5/41)<br>OCA2 without freckles 9.8% (4/41)<br>OCA2 or OCA3 7.3% (3/41)<br>OCA2 or BOCA or OCA3 61% (25/41)<br>OCA2 or BOCA 9.8% (4/41) |
| <b>Social discrimination</b>                             | 53.7% (22/41) mild<br>12.2% (5/41) severe<br>34.1% (14/41) none                                                                                                  |

Based on phenotype, suspected OCA subtypes were categorized as follows: OCA2 with freckles (12.2%, 5/41), OCA2 without freckles (9.8%, 4/41), OCA2 or OCA3 (7.3%, 3/41), OCA2/BOCA/OCA3 (61%, 25/41), and OCA2 or BOCA (9.8%, 4/41) (Table 1).

Social discrimination emerged as a critical issue. Overall, 65.9% (27/41) reported experiences of discrimination: 53.7% (22/41) described mild forms (at school, work, or within the community), and 12.2% (5/41) reported severe episodes of physical or psychological abuse.

In contrast, 34.1% (14/41) reported no discrimination, and this subgroup was exclusively from Diego Suarez, suggesting a more supportive or educated urban environment.

Regarding photosensitivity and photodamage, 90.2% (37/41) reported previous sunburns, highlighting the lack of photoprotection. At the initial visit, 53.7% (22/41) presented with severe solar erythema, 39.0% (16/41) with mild erythema and 7.3% (3/41) showed no signs of sunburn. Concerning clothing, 22.0% (9/41) wore covering attire, while 78.0% (32/41) wore non-covering clothing.

Sun protection strategies were generally inadequate: 39% of patients (16/41) reported using protective clothing, 7.3% (3/41) reported sunscreen use (none consistently), and 19.5% (8/41) avoided midday sun exposure. Nearly half (43.9%, 18/41) reported no sun protection practices at all (Table 1).

All participants received an individualized education during the initial visit, supported by interpreters and culturally adapted materials. These materials emphasized the importance of sun protection to prevent sunburn and skin cancer, the correct and consistent use of sunscreen, and practical behaviors such as wearing wide-brimmed hats and long-sleeved clothing and seeking shade during peak sunlight hours. To ensure cultural relevance, the materials were translated into Malagasy, simplified for low-literacy contexts, and supplemented with illustrations and examples relatable to the local culture (e.g., traditional clothing styles, daily outdoor activities). They were provided with SPF 50+ sunscreen, wide-brimmed hats, sunglasses, and donated long-sleeved clothing. Literate participants also received bilingual leaflets in Malagasy and French.

A total of 48.8% (20/41) attended monthly follow-up visits throughout the six-month period. Among completers ( $n = 20$ ), paired analysis showed marked improvements in photoprotection. Sunburn prevalence decreased from 19/20 (95.0%) at baseline to 2/20 (10.0%) at follow-up (exact McNemar  $p < 0.0001$ ). Regular sunscreen use increased from 0/20 (0.0%) to 20/20 (100.0%) ( $p < 0.0001$ ). Use of protective clothing rose from 7/20 (35.0%) to 16/20 (80.0%) ( $p = 0.0039$ ), and adoption of behavioral strategies rose from 3/20 (15.0%) to 10/20 (50.0%) ( $p = 0.0156$ ). Reporting of no protection declined from 11/20 (55.0%) to 0/20 (0.0%) ( $p = 0.0010$ ). All comparisons were statistically significant (Table 2), and none of the completers remained without any form of sun protection.

**Table 2.** Changes in sun protection behaviors from baseline to 6 months follow-up. Values are presented as percentages with absolute numbers in parentheses. “No protection” refers to participants reporting no use of sunscreen, protective clothing, or behavioral strategies. Statistical significance was assessed using the exact McNemar test.

| Behavior              | Baseline    | Follow-Up    | $p$ -Value with McNemar |
|-----------------------|-------------|--------------|-------------------------|
| Sunburn prevalence    | 95% (19/20) | 10% (2/20)   | $p < 0.0001$            |
| Regular sunscreen use | 0% (0/20)   | 100% (20/20) | $p < 0.00001$           |
| Protective clothing   | 35% (7/20)  | 80% (16/20)  | $p = 0.0039$            |
| Behavioral strategies | 15% (3/20)  | 50% (10/20)  | $p = 0.0156$            |
| No protection         | 55% (11/20) | 0.0% (0/20)  | $p = 0.001$             |

These results demonstrate a significant and consistent improvement in photoprotective behaviors following the structured intervention program, despite the small sample size inherent to this rare disease.

#### 4. Discussion

This observational study provides novel data on the clinical features, sun-related health burden, and social challenges of individuals with OCA in northern Madagascar, a population for which epidemiological evidence has been largely lacking. Our findings

highlight the dual vulnerability of this group, encompassing both biomedical and socio-cultural dimensions.

The dermatological profile of our cohort was consistent with reports from sub-Saharan Africa, with a high prevalence of actinic keratoses and a striking frequency of skin cancer already in early adulthood [4,5,7,12,16]. Lookingbill et al. [16] reported that actinic keratoses were present in 91% of OCA subjects over the age of 20 years in rural Tanzania, reaching 100% by age 30. Similarly, a study conducted at Kisangani Clinic in the Democratic Republic of Congo [5] found actinic keratoses in 100% of participants; however, the study population was on average older than in our cohort (49% of our patients were younger than 18 years, compared to only 23% in the Congolese study). In the same study, the prevalence of skin cancer was 29.8% versus 5% in our cohort, likely reflecting the older age distribution. Furthermore, in a large study of 350 individuals with albinism in Dar-es-Salaam, Tanzania [12], actinic keratoses were documented in all participants over the age of 20. Compared to studies from Tanzania and the Democratic Republic of Congo [5], the prevalence of precancerous and malignant lesions in our cohort was similarly high, supporting the urgent need for targeted skin cancer prevention programs across the region.

Baseline data confirmed extremely poor photoprotection practices, with nearly half of the participants reporting no protective measures at all. This reflects barriers repeatedly described in African settings, including limited awareness, scarce availability of sunscreen, high costs, and cultural preferences for non-covering clothing [17,18]. Importantly, our structured intervention program achieved significant improvements: sunburn prevalence dropped from 90% to 10%, while regular sunscreen use and protective clothing adoption rose sharply. Several challenges were encountered, mainly related to local cultural beliefs and initial mistrust among patients with albinism. These were addressed through continuous education and community engagement. From September 2024, the participation of an albino university staff member helped increase trust and encouraged more patients to join the program. Although follow-up was limited to six months and only 48% of patients were retained, these results demonstrate that community-based, low-cost interventions can effectively induce behavioral change in high-risk populations.

Two-thirds of participants reported experiences of social discrimination, ranging from exclusion to physical and psychological abuse. This is consistent with previous studies from Tanzania, Malawi, and South Africa, where myths and misconceptions about albinism contribute to profound human rights violations [1,13]. Interestingly, patients living in the urban setting of Diego Suarez reported fewer discriminatory experiences, suggesting that education, awareness campaigns, and community support may mitigate stigma.

Although genetic testing was not available, phenotypic stratification suggested that most patients fell within the OCA2, BOCA, or OCA3 subgroups, in line with prior genotype–phenotype correlations reported in African populations. While clinical classification is imperfect, it remains a useful tool in resource-limited settings until molecular diagnostic capacity becomes available.

This study has several limitations. The small sample size and limited follow-up restrict generalizability and preclude long-term outcome assessment. Loss to follow-up further reduced statistical power; however, this was largely attributable to geographical barriers, as only participants from the DIANA region could easily reach Diego Suarez for follow-up visits. Participants from the SAVA region expressed willingness to attend, but distance and transportation difficulties prevented their participation. Finally, the lack of genetic testing prevented a definitive molecular diagnosis.

Despite these limitations, this study demonstrates both the magnitude of dermatological morbidity and the feasibility of community-based interventions in people with albinism in Madagascar. Scaling up such programs—through partnerships with local

health authorities, NGOs, and patient associations—could substantially reduce the burden of sun-induced disease and improve quality of life. Future work should include longitudinal monitoring, integration of ophthalmologic and psychosocial care, and efforts to expand access to molecular diagnostics.

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**Informed Consent Statement:** Informed consent was obtained from all participants or patients’ parents/guardians in the case of minors. For illiterate participants study information was read aloud in Malagasy or French by the interpreter, and consent was documented by thumbprint or mark in the presence of a witness.

**Data Availability Statement:** The original contributions presented in this study are included in the article. Further inquiries can be directed to the corresponding author.

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## Abbreviations

The following abbreviations are used in this manuscript:

|      |                               |
|------|-------------------------------|
| OCA  | Oculocutaneous albinism       |
| UV   | Ultraviolet                   |
| AKs  | Actinic keratoses             |
| SCC  | Squamous cell carcinoma       |
| BCC  | Basal cell carcinoma          |
| CMM  | Cutaneous malignant melanoma  |
| BOCA | Brown oculocutaneous albinism |

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