

## Albinism in the Democratic Republic of Congo: Epidemiological and Clinical Aspects in Mbuji-Mayi, Kasai Oriental

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### Abstract

### Original Research Article

Albinism is a genetic mutation that affects the production of melanin in the skin, hair and eyes, making people with albinism vulnerable to ultraviolet radiation, with a greater susceptibility to skin cancer. In the Democratic Republic of Congo (DRC) access to care for people with albinism remains difficult. The province of Kasai Oriental is reported to have a large number of people with albinism and no local documentation has been provided to date. The objective of this pilot study was to describe the epidemiological and clinical aspects of albinism, which will enable the development of programs to prevent albinism-related complications in Mbuji-Mayi, Kasai Oriental. This is a cross-sectional and descriptive study conducted in the town of Mbuji-Mayi, capital of the province of Kasai Oriental, among people with albinism over a period of 7 days. Dermatological and ophthalmological diagnoses were essentially clinical. A total of 77 people with albinism of both sexes, most of them under 30 years of age, from all over the town were seen in consultations. Regarding albinism-related diseases, we observed basal cell and squamous cell carcinomas in identical proportions. In general, 78% of the people with albinism seen were at their first medical visit for a dermatological examination and only 4.2% of the people with albinism applied sunscreen. The present study revealed a not negligible frequency of albinism with a presence of pre-cancerous and cancerous lesions and a low rate of dermatological medical visits. This confirms the difficult access to medical care and information, justifying this very low photoprotection.

**Keywords:** Albinism, epidemiological clinics, photoprotection.

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## INTRODUCTION

Albinism is a genetic mutation caused by alterations in at least 21 genes identified to date, which affect melanin production in the skin, hair and eyes and is characterized clinically by widespread cutaneous hypopigmentation, photosensitivity, nystagmus, strabismus and reduced vision [1-3]. This melanin defect makes individuals vulnerable to ultraviolet (UV) radiation with an increased susceptibility to skin cancer [3]. The diagnosis of albinism is clinical and based on dermatological and physical examination (skin and eye depigmentation) [1]. Electrophysiological studies and imaging (Optical Coherence Tomography) complete the diagnosis [4]. There are different forms of albinism

classified into subtypes, and confirmation is based on the identification of the responsible gene [1, 3, 5].

Albinism affects on average 1 in 17,000 people worldwide [1, 5]. In sub-Saharan Africa, the prevalence of albinism is estimated to be in the range of 1 in 2000 to 5000 [5-7]. In the Democratic Republic of Congo (DRC), demographic data on people with albinism are limited to hospital studies [8, 9]. Access to care remains difficult, due in particular to extreme poverty, population displacement, the absence of specialists in dermatology in several provinces and the lack of an organization to support albinism [8]. In a study on skin lesions observed in oculocutaneous albinism in 200 patients in Kinshasa in 2016, Kakiése *et al.*, show that Kasai Oriental province was the area of origin of most

people with albinism with 28% or 57 out of 200 patients in the study [9]. There is too little data on albinism in Mbuji mayi in Kasai Oriental in DRC [11]. This pilot study was conducted in the town of Mbuji mayi with the objective of describing the epidemiological and clinical aspects of albinism, which will allow the development of programs to prevent complications related to albinism in Mbuji mayi, East Kasai.

## METHOD

**Background to the study:** This study took place in the town of Mbuji mayi, the capital of the province of Kasai Oriental, located in central- eastern DRC (Figure 1). It is subdivided into five communes (Muya, Dibindi, Kanshi, Diulu and Bipemba). It is the third largest city in the country in terms of population. The population is estimated at 2,525,263 in 2021 (Mbuji mayi 2021 population data), covering an area of 135.12 km<sup>2</sup> and corresponding to a population density of 12,441 inhabitants/km<sup>2</sup> [11].



**Figure 1: Location of the town of Mbuji mayi on the map of the Democratic Republic of Congo (DRC) [11]**

**Type of study and inclusion criteria:** This is a descriptive prospective cross-sectional study over a period of 7 days, from 8 May to 14 May 2022, among people with albinism, our study population. We included in the study all AAPs who were aware of and came alone or accompanied to the consultation during the study period. People with albinism who were unable to provide all the information in the interview were excluded.

**Study variables and operational definitions:** The socio-demographic variables were: age, sex, occupation and commune of origin). Clinical variables: family history of albinism, number of medical visits for a dermatological examination, notion of sun protection (physical: hats and long-sleeved clothes and chemical: anti-sun cream) and the clinical dermatological and ophthalmological manifestations observed.

**Table 1: Operational definitions and diagnostic criteria for albinism**

| <b>Criteria for the diagnosis of albinism</b>                     | <b>depigmentation of the skin (including hair) and the eye (with visual impairment)</b>   |
|---|---|
| Dermatological conditions observed in AAPs (people with albinism) | These are: Basal cell carcinoma, squamous cell carcinoma, actinic keratosis, ephelides, lentigines, nevi, cheilitis and undifferentiated tumors.  |
| The profession  | Any professional occupation including studies   |
| Sun protection  | Physical: wearing long-sleeved clothes, wide- brimmed hats<br>Chemical: regular use of an SPF 50 <sup>+</sup> cream to protect against ultraviolet A and B rays in exposed areas not covered by clothing. |
| Sunscreen cream   | Any SPF 50 cream <sup>+</sup> (protecting against ultraviolet A and B rays).  |
| Ophthalmological disorders  | Strabismus, nystagmus and ectropion.  |

In this study, the dermatological and ophthalmological diagnoses were essentially clinical on the basis of a predefined criterion (see above) and we were interested only in lesions frequently associated

with albinism. Other dermatoses were not taken into account.

**Data collection:** In conjunction with World Albinism Awareness Day (13/06) each year [12], an awareness- raising campaign was conducted for 2 months (from 8 March to 8 May 2022), which ended with a series of free consultations for people with albinism, organized at the Mbujimayi pediatric clinic. The awareness-raising was made possible with the support of the non-governmental organization (NGO) Solidarité des albinos du Kasai oriental (SAKOR). The consultations were carried out by a group of Spanish dermatologists in collaboration with Congolese doctors. **Ethical approval:** All people with albinism or legal guardians of minor people with albinism provided written informed consent to participate in the study. The study protocol had been reviewed and approved by the Ethics Committee of the Faculty of Medicine of the University of Mbujimayi (Ref: 012/VD-RSCU/Fac-Med/UM/DMT/2019). The study was conducted in accordance with the principles of the Declaration of Helsinki (2013 version) [14]. The purpose and procedures of the study were explained to participants and legal respondents. Inclusion was risk-free for people with albinism. Participants' anonymity was guaranteed and no personal details were recorded. The statistical analysis of the collected data was done using EPI Info software version 7.1.3.3. The data, depending on the case, are represented by the average or the frequency.

## RESULTS

A total of 77 people with albinism were seen in consultations during the study period. Socio-demographic characteristics of people with albinism: Of the 77 people consulted, 50 (64.9%) were female, with a M/F sex ratio of 0.54. Patients under 25 years of age predominated with 76.7% (n=59). The mean age of our patients was  $21.7 \pm 13.7$  with extremes of 2 and 72 years. The patients came from all 5 communes of the city: Dibindi commune was more represented with 26 cases or 33.8%, followed by Bipemba with 16 cases (20.8%) and Diulu commune had 14 patients or 18.2%. Thirty-four patients (44.2%) had no professional occupation and 31 patients (40.2%) were students and pupils, other professions were present: farmer (n=2), teacher (n=2), shopkeeper (n=4), nurse (n=2), seamstress (n=2) (Table 2).

Characteristics according to family history of albinism and number of medical visits: No family history of albinism was noted in 43 patients (55.8%) and 34 patients (44.2%) acknowledged having 1 or more PAAs in their family. This consultation was a first medical visit for a skin examination for 78% (n=60) of patients, a second for 17 patients (22%) and no patient had already had more than 2 medical visits (Figure 1).

**Notion of sun protection:** Sun exposure was recognized by 71 patients (92.2%) of whom 40.8% (n=29) practiced regular physical sun protection and only 4.2% (n=3) practiced chemical sun protection in addition (application of sunscreen) (Table 3).

**Frequency of dermatoses related to albinism:** During the period of our study, our patients were diagnosed with: 3(3.9%) cases of basal cell carcinoma, 3(3.9%) cases of squamous cell carcinoma, 3(3.9%) cases of undifferentiated tumors, 24(31.2%) patients with actinic keratosis, 52 (67.5%) cases of ephelides, nevus and lentigines; and 8 cases of actinic cheilitis (Table 4). The cancers are located on the head and upper limbs with 3 cases for both sexes. **Frequency of ophthalmological abnormalities:** Concerning ophthalmological damage, we noted 20 cases of strabismus, 67 cases of nystagmus and 2 patients with ectropion (Table 5).

**Table 2: Socio-demographic distribution of people with albinism**

|                          | Workforce | Percentage (%) |
|--------------------------|-----------|----------------|
| <b>Gender</b>            |           |                |
| Female                   | 50        | 64,9           |
| Male                     | 27        | 35,1           |
| <b>Age group</b>         |           |                |
| ≤5                       | 6         | 7,8            |
| ]5 - 15]                 | 19        | 24,7           |
| ]15 - 25]                | 34        | 44,2           |
| ]25 - 35]                | 7         | 9,1            |
| ]35 - 40]                | 3         | 3,9            |
| >40                      | 8         | 10,4           |
| <b>Commune of origin</b> |           |                |
| Bipemba                  | 16        | 20,8           |
| Dibindi                  | 26        | 33,8           |
| Diulu                    | 14        | 18,2           |
| Kanshi                   | 7         | 9              |
| Muya                     | 14        | 18,2           |
| <b>Profession</b>        |           |                |
| Agriculture              | 2         | 2,6            |
| No                       | 34        | 44,2           |
| Trade                    | 4         | 5,2            |
| Seamstress               | 2         | 2,6            |
| Teacher                  | 2         | 2,6            |
| Student and pupil        | 31        | 40,2           |
| Nurse                    | 2         | 2,6            |

Females predominated in our study with 65% of cases. The age range between 15 and 25 years was predominant in our study with 44.2% of the cases. The average age was  $21.7 \pm 13.7$ . The commune of Dibindi was more represented with 33.8%. Patients without any professional occupation predominated with 44.2% followed by students with 40.2%.

**Table 3: Distribution according to the practice of photoprotection (n=71)**

| Photo protection | N  | %    |
|------------------|----|------|
| <b>Physics</b>   |    |      |
| Yes              | 29 | 40,8 |
| No               | 42 | 59,2 |
| <b>Chemical</b>  |    |      |
| Yes              | 3  | 4,2  |
| No               | 68 | 95,8 |

Patients without regular physical protection predominated with 59.2% of the cases and those practicing chemical protection in addition (application of sunscreen) represented 4.2%.

**Table 4: Distribution according to albinism-related dermatoses (n=77)**

| Dermatoses                          | N  | %    |
|-------------------------------------|----|------|
| <b>CBC</b>                          |    |      |
| Yes                                 | 3  | 3,9  |
| No                                  | 74 | 96,1 |
| <b>EC</b>                           |    |      |
| Yes                                 | 3  | 3,9  |
| No                                  | 74 | 96,1 |
| <b>Undifferentiated tumors</b>      |    |      |
| Yes                                 | 3  | 3,9  |
| No                                  | 74 | 96,1 |
| <b>Actinic Keratosis</b>            |    |      |
| Yes                                 | 24 | 31,2 |
| No                                  | 53 | 68,8 |
| <b>Ephelides, Lentigines, Nevus</b> |    |      |
| Yes                                 | 52 | 67,5 |
| No                                  | 25 | 32,5 |
| <b>Actinic cheilitis</b>            |    |      |
| Yes                                 | 8  | 10,4 |
| No                                  | 69 | 89,6 |

BCC: basal cell carcinoma EC: squamous cell carcinoma

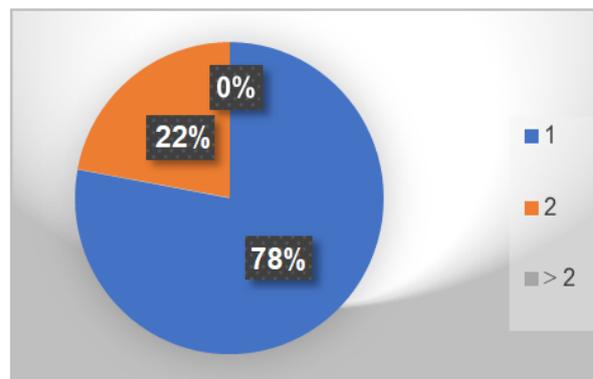
Basal cell carcinoma and squamous cell carcinoma are represented with 3.9% cases respectively and actinic keratoses with 31.2% cases.

**Table 5: Distribution according to ophthalmological anomalies (n=77)**

| Anomalies         | N  | %    |
|-------------------|----|------|
| <b>Strabismus</b> |    |      |
| Yes               | 20 | 26,0 |
| No                | 57 | 74,0 |
| <b>Ectropion</b>  |    |      |
| Yes               | 2  | 2,6  |
| No                | 75 | 97,4 |
| <b>Nystagmus</b>  |    |      |
| Yes               | 67 | 87,0 |
| No                | 10 | 13,0 |

Patients with nystagmus accounted for 87% of cases and those with strabismus accounted for 26% of cases.

Patients with only one medical visit predominated in our study with 78% of cases.

**Figure 1: Distribution according to the number of medical visits for dermatological examination**

## DISCUSSION

The survey aimed to determine the epidemiological and clinical aspects of albinism in Mbujimayi. A total of 77 people with albinism (PAA) of both sexes, mostly young and without occupations, some with carcinomas, were enumerated. This participation, whose recruitment was done in 7 days and on a single site, seems high compared to Koudoukpo C *et al.*, in Benin who recruited 99 patients between May and August 2020 from 4 associations, whose recruitment was done on two sites [15]. Our frequency is possibly biased, particularly because some people with albinism living at long distances did not come to the consultations.

Females predominated with 64.9% of cases. The gender difference in the epidemiology of albinism varies between authors [10, 15-17]. The case of Koudoukpo C *et al.*, who found 63.53% of patients to be male [15]. Our results are similar to those of Konate I *et al.*, in Burkina-Faso who found that 65% of patients were female [16]. In our study, the majority of patients were under 25 years of age with an average of 21.7 years. Our results are similar to those of Mouhari-Toure A *et al.*, in Togo with also a mean age of 21.7 years [18]. Several authors have found that many patients were under 30 years of age [19-21] and link this situation to the reduced life expectancy in people with albinism.

Regarding occupation, 44.2% of our patients were unemployed and 40.2% were students. Our results are similar to those of Kakiese *et al.*, who found 41% of the people with albinism were unemployed [10]. Koudoukpo C *et al.*, found that pupils and students were the most represented (38.38%) and explained that this would be due to the fact that their study was done in an urban environment [15]. This is similar to our case, as Mbujimayi is an urban setting.

Only 44.2% of cases had a family history of albinism. This situation is close to that observed in Togo by Mouhari- Toure A *et al.*, who found a family history in 37.9%. This same team found a notion of consanguinity [18] which was cited in Kinshasa [10] but not demonstrated in this study.

In 78% of the cases, our patients were only at their first dermatological consultation. These results are due to the fact that in the Democratic Republic of Congo, people with albinism have difficult access to care due to the absence of specialists in dermatology in several provinces of the country [9] This is the case of Mbujimayi which has no dermatology service. Normally people with albinism must be followed up annually by their doctor [22] or at least twice a year [23].

Photoprotection is limited to physical protection (40.8%) (hats and long-sleeved clothing). Sunscreen is not widely used (4.2%), yet effective photoprotection from a young age can limit the development of skin carcinomas [3]. Our results are inferior to those of Koudoukpo C *et al.*, who found that 62.63% had appropriate clothing habits (long-sleeved clothes) and 12.12% used sun cream [15, 8]. Despite this difference, photoprotection remains insufficient in general [24]. This demonstrates the lack of education or information [25]. Our survey found 67.5% of patients with ephelides, nevi and lentigines and 31.2% with actinic keratoses. Some patients were found to have carcinomas. Our results are similar to those of Koudoukpo C *et al.*, who observed ephelides as the main dermatosis with 56.57%, followed by actinic keratosis (24.24%) [15]. Our results do not show a difference between carcinomas and are similar to those of Ramos AN *et al.*, who observed 8 patients with basal cell vs 7 with squamous cell [26] although in the literature squamous cell carcinoma is the most frequent [3]. Our results confirm those of Gassama *et al.*, who found that skin cancer is frequent and accessible by sight and therefore by clinical examination [27]. Our results on ophthalmic expectations are similar to those observed by Konate *et al.*, with 100% of people with albinism examined presenting at least one eye condition [16]. This is normal as these visual disorders are constitutional [8, 16]. Our study is limited to Mbujimayi, which makes it impossible to generalize given our sample. Our diagnoses were only clinical due to technical, time and financial issues. This constitutes the limitations of our study. Despite these limitations, this pilot study has provided a view and literature on albinism in Mbujimayi. Further work will allow us to explore in greater depth the aspects not covered by this study.

## CONCLUSION

This pilot study in Kasai Oriental shows the epidemiological and clinical aspects of albinism to proportions that deserve special attention. Access to

dermatological care and medical education remains insufficient. We believe that regular thematic consultations in dermatology will improve the management of people with albinism.

**Conflicts of interest:** The authors declare no conflict of interest.

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