

Case Report

Sun Safety Struggles Among Children with Xeroderma Pigmentosum in a Tropical Low-Income Country

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Abstract

Xeroderma pigmentosum (XP) is a rare genodermatosis characterized by increased sensitivity to ultraviolet radiation, leading to severe skin manifestations and a higher risk of early-onset malignancies. Previous studies from temperate climate countries with sound economic levels showed adequate photoprotection compliance among pediatric XP patients. However, no studies have assessed photoprotection compliance among children with XP living in tropical and low-economic settings. This article reports a low photoprotection compliance of three pediatric XP patients residing in Indonesia, a tropical low-income country. The three patients began experiencing their first symptoms in the first year of life with a gap of 1-3 years until they were diagnosed with XP by dermatologists. Photoprotection measures were promptly initiated. However, challenges related to the hot climate and low economic status led to lower levels of photoprotection compliance. Ultimately, the three patients developed UV-associated skin malignancy at early ages. This report underscores the challenges of maintaining a favorable prognosis for XP patients in low-income tropical countries.



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Keywords

Genodermatosis; photoprotection compliance; xeroderma pigmentosum

1. Introduction

Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis clinically characterized by a marked sensitivity to ultraviolet (UV) radiation and a significant likelihood of developing various skin malignancies. The incidence rate is anticipated to range from 1 in 250,000 to 1 in 1,000,000 in North America and Europe [1]. It occurs as an expression of a gene mutation involved explicitly in DNA repair after exposure to ultraviolet radiation (UVR). Depending on the gene type affected, several XP complementation categories have been identified (XPA-XPG and XPV) [2]. Generally, patients with XP develop extensive freckles in sun-exposed areas before the age of 2 years. Over time, the skin goes through premature aging, characterized by progressive xerosis, atrophy, wrinkling, lentigos, poikiloderma, and early onset of skin and ocular malignancies. Due to these reasons, sun avoidance stands out as the primary management strategy for all patients. At the same time, other approaches, such as treating actinic keratosis, poikiloderma, skin cancers, and genetic and psychological counseling, may vary depending on individual circumstances [2]. Limiting UV exposure by providing unwavering, permanent photoprotection is paramount for XP patients, especially young ones. Patients must avoid sunlight to the utmost extent, protect their faces with UVR-protective transparent film visors, and cover the rest of their bodies with clothing, hats, gloves, and highly protective sunscreen [3].

Few studies have assessed specific photoprotection compliance in XP patient populations. Walburn et al. conducted one study that effectively assessed this aspect in 2019 [3]. The study evaluated the level of photoprotection in pediatric and adult patients with XP in the United Kingdom, France, Germany, and the United States. They utilized self-reporting measures that were validated against objective measures [4]. The self-reporting measures employed a five-point ordinal scale with 1 being the lowest and 5 being the highest for each photoprotection category, including photoprotection for face and body, and compliance in avoiding going outside. Scores below 4 indicated inadequate compliance (refer to the cited reference for the complete protocol development and validation process) [3, 4]. The study showed that adult patients had poorer photoprotection levels compared to pediatric patients or the cared-for sample, both for face (2.89 ± 1.22 vs. 4.19 ± 0.73) and body (3.70 ± 1.10 vs. 4.65 ± 0.63) ($p < 0.001$). The results indicate that children cared for by a caregiver tended to have relatively good levels of photoprotection compliance.

Unfortunately, no study has assessed the level of photoprotection compliance among patients with XP in tropical countries, particularly pediatric patients, despite the high UV index throughout the year and its more significant impact on XP progression in such regions. Therefore, the authors aimed to report the photoprotection compliance of pediatric XP patients treated at our tertiary hospital in Yogyakarta, Indonesia. Due to the rarity of XP, only three pediatric patients could be assessed. The three patients' caregivers were interviewed using the same self-reporting measures from the previous study. Factors contributing to photoprotection compliance were also evaluated.

2. Case Report

2.1 Patient 1

A 5-year-old boy presented with extensive pigmentation covering sun-exposed areas of the body. The caregivers explained that the freckling symptoms began when the patient was 1 year old, and he was diagnosed with XP at 2 by the dermatologist. The dermatologist informed the caregivers about the sensitivity to UVR and advised them to practice strict photoprotection, including sunscreen. However, the patient refused to use sunscreen or other photoprotection measures due to the oily sensations of sunscreens and the sticky and hot feeling when wearing long-sleeved clothes. The family also found the expenses of purchasing sunscreen for daily use burdensome. Unfortunately, at age 4, the patient developed a large mass on the nose. The patient underwent a wide excision of the mass by a plastic surgeon, and the histopathological examination confirmed a squamous cell carcinoma (SCC). The patient was subsequently referred to us for general dermatological care.

2.2 Patient 2

A 4-year-old boy presented with a similar extensive pigmentation pattern as Patient 1. The patient was referred by an ophthalmologist for general dermatological care after undergoing a wide excision of a sizeable ulcerative mass from his left conjunctiva, which is histopathologically evident for conjunctival SCC. According to his parents, skin pigmentation symptoms started when the patient was 1 year old and was diagnosed with XP at 2 by the dermatologist. The patient also experienced a developmental delay. The caregivers received all the required information regarding the risk factors and the importance of photoprotection. However, like Patient 1, he refused to comply with photoprotection due to hot weather and uncomfortable feeling after using sunscreen during the day. The cost required to purchase sunscreen for long-term use was also deemed burdensome. Regrettably, at the age of 4, the patient developed conjunctival SCC associated with UVR.




2.3 Patient 3

A five-year-old girl presented with similar pigmentary changes as seen in previous patients and also with developmental delay. According to her parents, the pigmentary changes began when she was 1 year old. However, there was a three-year gap until the parents sought medical professional advice. The patient was diagnosed with XP at 4 and started photoprotection measures promptly. However, the parents faced challenges in photoprotection because the child refused to use sunscreen due to the discomfort of applying sunscreen and using photoprotective clothes in hot weather. Unfortunately, at the age of 5, a hyperkeratotic mass developed on her right ear lobe. The patient underwent a wide excision of the mass on the right ear lobe by a plastic surgeon, and the histopathological examination confirmed an SCC. She was then referred to us for general dermatological care.

3. Photoprotection Compliance Score

Sun-damage symptoms started to develop in the first year of life for three patients, and there was a delay of 1 to 3 years between the onset of symptoms and their physicians' initial XP diagnosis. All patients started photoprotection promptly after being explained about their sensitivity to UV radiation. However, the interview with the caregivers regarding the photoprotection compliance level, using the same self-reporting measures from the previous study, showed disheartening results. The three children showed a median score of 2.6 for face photoprotection, 3.0 for body photoprotection, and 2.0 for compliance in avoiding going outside (scores below 4 indicate inadequate photoprotection) (Table 1).

Table 1 Demographic features, clinical pictures, and photoprotection behaviors in three pediatric XP patients in a tropical low-income country.

	Patient 1	Patient 2	Patient 3
			
Demographic features			
Gender	Male	Male	Female
Current age	5 y.o.	4 y.o.	5 y.o.
Age of symptoms onset	1 y.o.	1 y.o.	1 y.o.
Age at diagnosis	2 y.o.	2 y.o.	4 y.o.
Age at which started photoprotection	2 y.o.	2 y.o.	4 y.o.
Total family income per month	400-500 USD	300-400 USD	300-400 USD
Clinical features			
Abnormal sunburn	No	No	Yes
Pigmented sun-damage changes (freckles, lentigo, hypopigmentation, poikiloderma)	Yes	Yes	Yes
Skin cancer	Yes (SCC)	Yes (SCC)	Yes (SCC)
Ocular manifestation	No	Yes	Yes
Neurology symptoms	No	Yes	Yes

Photoprotection behaviors score			
(lowest score 1, highest score 5, minimum score 4 for adequate photoprotection)			
Face photoprotection	3.0	2.6	2.0
Body photoprotection	3.0	3.0	2.0
Avoid going outside	2.0	2.0	2.0

4. Discussion

Indonesia is a tropical country with high humidity (82-85%) and hot temperatures all year round, with an average annual temperature of 27°C that can rise to 32°C during the day [5]. This climate is markedly different from the countries where the previous study was conducted, which experience four seasons: the United Kingdom, France, Germany, and the United States. The temperatures in these countries vary throughout the seasons, with average annual temperatures in the last years before 2022 being only 9.1°C, 14.6°C, 10.6°C, and 13.9°C, respectively [5]. The climate disparity poses a much more uncomfortable experience in using photoprotective materials in tropical compared to non-tropical countries, explaining the lower photoprotection compliance of our patients. According to the parents, during the first 1-2 years of their child's life, they can still be convinced to refrain from playing outside during the daytime. However, as the child grows older and has peers around the neighborhood, they constantly cry and complain they want to go out. The parents want to provide adequate photoprotection, but the hot weather conditions in Indonesia make the child reject sunscreen during the daytime due to the sticky and oily sensation it leaves on their skin. The children also refuse to wear covered clothing due to the hot weather.

Besides the hot climate, Indonesia is a developing country with a low level of economic development. The majority of the population still has a low family income per month. Since national health insurance does not cover photoprotective materials, the families must bear additional expenses. The highest cost factor in caring for XP patients is the lifelong use of sunscreen. The ideal amount of effective sunscreen used to obtain protection from the SPF stated on the label packaging is 2 mg/cm² [6]. The estimated body surface area of a 5-year-old child is around 7400 cm² [7]. Therefore, the amount of sunscreen a 5-year-old XP patient requires to optimally protect sun-exposed regions (head, neck, and extremities) that cover 69% of the body surface area is 14.8 grams. The Indonesian market's cheapest sunscreen lotion with SPF 30 and PA+++ (the minimum recommended photoprotection factor) is 3.5 USD for 100 grams. Based on this calculation, the cost for daily use with four re-applications will require 2 USD per day and 60 USD per month. Sunscreen as a consumable product is expensive for the three patients' families, reaching 12-20% of the total family income (300-500 USD per month).

In this report, all patients developed an early onset of UVR-associated skin malignancy. Failure to comply strictly with photoprotection may accelerate the progression. However, the type of XP complementation category may also influence this condition. Some complementation groups may predispose the patient to have early-onset skin malignancies, such as XPA (the most common group in Japan) and XPC (most common in the United States and Western Europe) [8]. Unfortunately, due to inaccessible facilities, none of the three patients reported in this article were tested for the complementation group. Therefore, more studies are needed to compare the influence of photoprotection compliance level and the XP complementation groups in developing early-onset malignancies.

5. Conclusion

Children with XP encounter unique challenges in low-income tropical countries, leading to lower levels of photoprotection compared to similar patients in developed countries with temperate climates. Innovative solutions would be beneficial to improve sun safety practices in this vulnerable population, such as creating more comfortable and affordable sunscreen formulations and establishing relaxed and sun-free playing environments for children. These measures would greatly help improve photoprotection compliance among pediatric XP patients and ultimately improve the quality of life for children with XP in low-income tropical countries.

Author Contributions

Yohanes Ridora was responsible for patients' data collection, drafted the case report manuscript, and reviewed and approved the final version for publication. Niken Trisnowati contributed to the interpretation of clinical data, assisted in drafting the manuscript, and reviewed and approved the final version for publication; Retno Danarti was responsible for providing oversight and guidance in case report preparation, assisting in interpreting clinical data, revising the manuscript critically for intellectual content, finalizing, and approving the manuscript for submission and publication.

Competing Interests

The authors have declared that no competing interests exist.

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