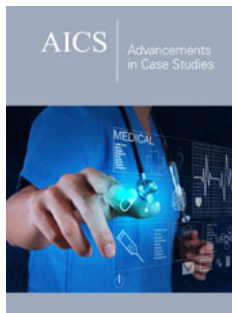


# Basal Cell Carcinoma in a Filipino with Oculocutaneous Albinism: A Case Report

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## Abstract

**Background:** Oculocutaneous albinism (OCA) is an autosomal recessive disorder characterized by defects in melanin synthesis that affect the skin, eyes, and hair. This melanin deficiency causes albino patients to be at a higher risk for sun-induced skin cancers. Herein, this report highlights the rare occurrence of basal cell carcinoma (BCC) in a Filipino with oculocutaneous albinism.

**Case:** A 25-year-old Filipino female presented with a 1-year history of a keratotic plaque on the neck. She had milky-white skin, silvery hair, and blue eyes at birth that gradually darkened with age with progressive photosensitivity and photophobia. No family history of albinism nor consanguineous marriage was noted. Physical examination revealed a fair-skinned patient with brown hair, gray-tinted irises, marked nystagmus, and decreased visual acuity. Clinical and genetic counseling was done leading to the diagnosis of OCA IB. In addition to this, an erythematous keratotic plaque on the left side of the neck was noted and upon dermoscopy, short fine telangiectasias, ulceration and yellow crusts were seen. Skin biopsy was done that showed tumor islands with atypical basaloid cells and a retraction artifact diagnostic of nodular BCC. She underwent Mohs micrographic surgery for her BCC, was recommended to follow-up regularly for screening, and was advised strict sun protection.

**Conclusion:** The hypopigmented skin of this patient with oculocutaneous albinism predisposed her to severe skin damage that led to the development of basal cell carcinoma. Due to melanin deficiency, it is recommended to closely observe, regularly follow-up, and advise strict sun protection as early as possible to prevent skin cancer in this population.

**Keywords:** Oculocutaneous albinism, basal cell carcinoma, squamous cell carcinoma, melanin, Nonmelanoma skin cancer, Mohs micrographic surgery, ultraviolet

**Abbreviations:** OCA: Oculocutaneous Albinism; BCC: Basal Cell Carcinoma; UV: Ultraviolet; NMSC: Nonmelanoma Skin Cancer; H&E: Hematoxylin & Eosin

## Introduction

Oculocutaneous albinism (OCA) is an autosomal recessive genetic disorder associated with reduced melanin biosynthesis. Clinical manifestations include hypopigmented skin, eyes, and hair, with visual disturbances such as impaired visual acuity, nystagmus, and photophobia [1]. Globally, OCA has an overall prevalence rate of approximately 1 in 17,000 with eight subtypes. Of these, the OCA1 subtype is the most common with milder mutations that allow residual tyrosinase activity. This makes affected individuals gain some pigment over time with slightly improved visual acuity [2].

Melanin is a photoprotective pigment that protects the skin from the harmful effects against ultraviolet (UV) radiation and oxidative damage to DNA [3]. The lack or absence of melanin predisposes skin to UV-induced damage, which includes sunburns, blisters, and actinic keratosis wherein sun-exposed areas located at the face, neck, and shoulders, are particularly prone to transforming into skin malignancies [1,2]. In albino individuals, they commonly develop squamous cell carcinoma (SCC) but other less frequently encountered

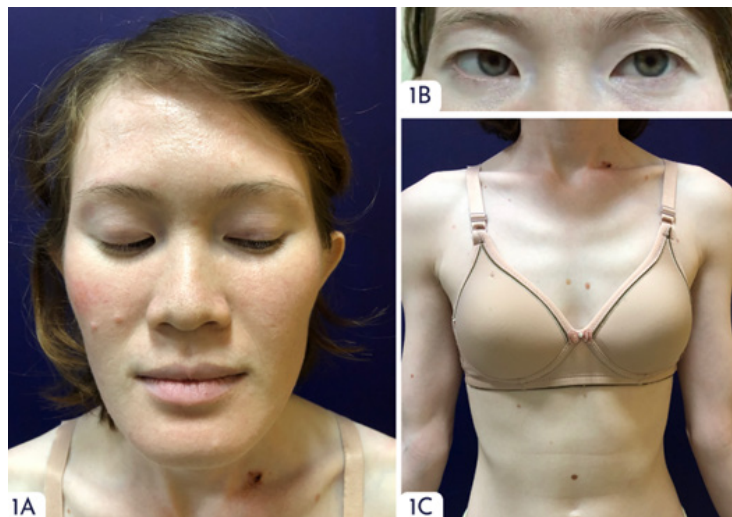
malignancies were basal cell carcinoma, dysplastic nevus, and melanoma [3]. In addition to this, many international studies have investigated the prevalence of nonmelanoma skin cancer (NMSC) such as BCC and SCC in patients with albinism. However, there were no studies of this nature in the Philippines which highlights the rare occurrence of basal cell carcinoma in a Filipino with oculocutaneous albinism. Persons of color have an increased morbidity and mortality associated with NMSC and this, in combination with the lack of resources or education devoted to skin cancer prevention in the Philippines, highlights the need for more emphasis on this issue [2]. Early diagnosis is critical to inform proper management, education, and well-being of OCA individuals with BCC but as this is not solely based on the skin, patients should also be referred to an ophthalmologist to monitor the patient's ocular and visual abnormalities [1,2,4].

To our knowledge, this is the first reported case of basal cell carcinoma in a Filipino with oculocutaneous albinism as a risk factor. We hope that it expands our understanding of BCC in patients with OCA and encourages public health initiatives to focus more on

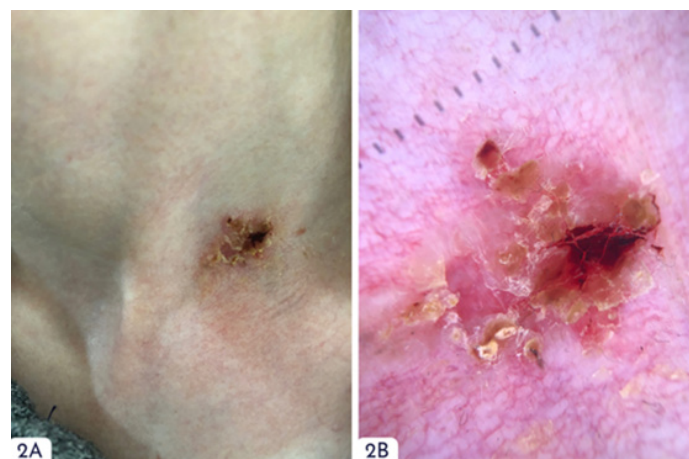
education and prevention [2,5].

### Case Presentation

A 25-year-old Filipino female presented to our clinic with a keratotic plaque on the left side of the neck. The lesion first appeared 1 year prior to consult as a small erythematous papule that progressively grew into an erythematous plaque topped with hemorrhagic crusts. She had milky-white skin, silvery hair, and blue eyes at birth that gradually darkened with age associated with progressive photosensitivity, photophobia, and nystagmus. No family history of albinism nor consanguineous marriage was noted. Physical examination revealed a fair-skinned patient with marked nystagmus and decreased visual acuity. Her hair was brown, while her irises had a light gray tint (Figures 1A to 1C). Clinical and genetic counseling was done which led to the diagnosis of OCA 1B. Multiple benign nevi throughout the body were evaluated but upon dermoscopy of the erythematous keratotic plaque on the neck, telangiectasias, ulceration and yellow crusts were appreciated (Figures 2A and 2B).



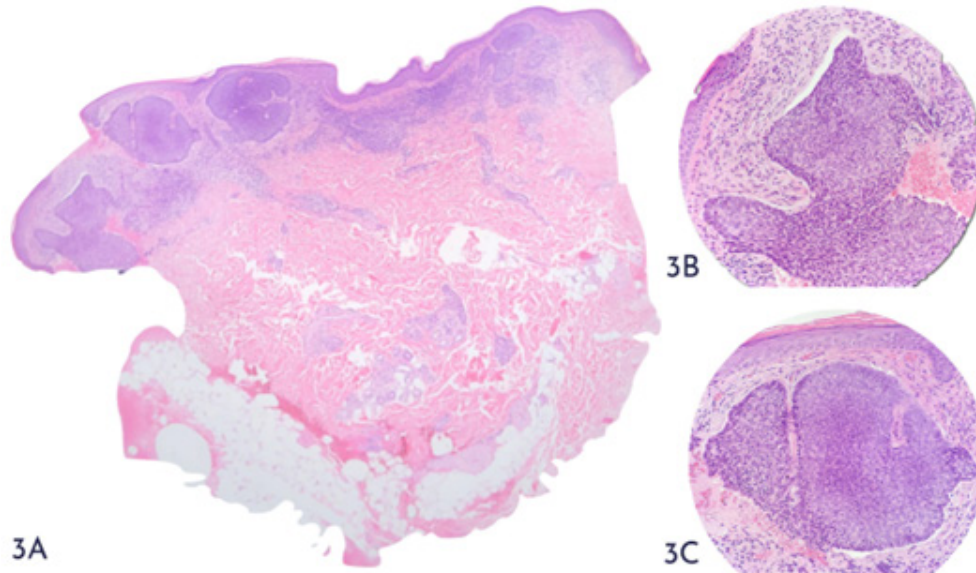
**Figures 1A to 1C:** Pigmentary dilution of skin with tan hair and gray-tinted irises characteristic of oculocutaneous albinism 1B. She had nystagmus but not visible here. (Photos taken with patient's consent)



**Figures 2A:** Clinical image of basal cell carcinoma at the left side of the neck. **2B:** On dermoscopy, telangiectasias, ulceration and yellow crusts were seen.

Biopsy was done on the lesion on the neck which showed tumor islands with atypical basaloid cells, peripheral palisading of tumor cells, and retraction artifacts in the epidermis with lymphocytic inflammatory stroma in the upper dermis diagnostic of nodular BCC (Figures 3A to 3C). She underwent stage I of Mohs micrographic

surgery and was prescribed with cefalexin and mupirocin cream post procedure. Due to the increased risk of recurrence, she was recommended to follow-up regularly for screening and advised strict sun protection.



**Figure 3:** Histopathology from a keratotic plaque on the left side of the neck. **A:** Hematoxylin & Eosin (H&E) staining of basal cell carcinoma at 40×. **B & C:** H&E at 400×. Note tumor islands with atypical basal cells, peripheral palisading, and retraction artifacts diagnostic of nodular basal cell carcinoma.

## Discussion

Oculocutaneous Albinism individuals have a 1000-fold risk for developing skin cancer with a well-established risk of recurrence than the general population due to the lack of melanin which is important against UV radiation and oxidative damage of DNA [2]. As such, the hypopigmented skin of this young OCA patient predisposed her to chronic and severe damage that led to the development of a malignant skin lesion. In a comparable study by Behera et al. (2017), our patient had features consistent with photodamage such as a malignant cutaneous tumor, affecting the photo-exposed area of the body with histopathology results consistent with basal cell carcinoma [3]. Although squamous cell carcinoma is the most common malignancy encountered in albinos, the occurrence of basal cell carcinoma cannot be ignored. Hence, close observation, regular follow-up, biopsy of the suspicious lesion, and the use of sun protection methods should be done at its earliest as skin cancer can be preventable in all patients, especially in those with albinism [3,6,7].

## Acknowledgements

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## Conflict of Interest

None

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