The New Ropanasuri Journal of Surgery

Volume 6 | Number 2 Article 7

12-30-2021

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Ahmad Kurnia
Division of Oncology Surgery, Department of Surgery, Faculty of Medicine Universitas Indonesia, Dr Cipto Mangunkusumo General Hospital, Jakarta, Indonesia, ahmadkurnia62@yahoo.com

Diani Kartini
Division of Oncology Surgery, Department of Surgery, Faculty of Medicine Universitas Indonesia, Dr Cipto Mangunkusumo General Hospital, Jakarta, Indonesia, d.kartini@gmail.com

Novinda Herwirastri
Training Program in Surgery, Department of Surgery, 3 Faculty of Medicine Universitas Indonesia, Dr Cipto Mangunkusumo General Hospital, Jakarta, vindahrw@gmail.com

Zoya M. Adyasa
Faculty of Medicine Universitas Indonesia, Dr Cipto Mangunkusumo General Hospital, Jakarta, Indoneisia, zooeyaaa@gmail.com

Recommended Citation

Kurnia, Ahmad; Kartini, Diani; Herwirastri, Novinda; and Adyasa, Zoya M. (2021) "Consequential Surgeries Limit Recurrence of Skin Malignancies in Xeroderma Pigmentosum: A Case Report," *The New Ropanasuri Journal of Surgery*: Vol. 6 : No. 2 , Article 7.
DOI: 10.7454/nrjs.v6i2.1111
Available at: https://scholarhub.ui.ac.id/nrjs/vol6/iss2/7

The New Ropanasuri Journal of Surgery
OPEN ACCESS
Published by Department of Surgery
Faculty of Medicine, Universitas Indonesia

Consequential Surgeries Limit Recurrence of Skin Malignancies in Xeroderma Pigmentosum: A Case Report

Ahmad Kurnia,[1] Novinda Herwirastri,[2] Diani Kartini,[1] Zoya M Adyasa[3]

1 Division of Oncology Surgery, 2 Training Program in Surgery, Department of Surgery, 3 Faculty of Medicine Universitas Indonesia, Dr Cipto Mangunkusumo General Hospital, Jakarta.

Corresponding author: ahmadkurnia62@yahoo.com Received: 8/Dec/2021 Accepted: 19/Dec/2021 Published: 30/Dec/2021
Website: https://scholarhub.ui.ac.id/nrjs/ DOI:10.7454/nrjs.v6i2.1111
Ahmad Kurnia https://orcid.org/0000-0001-8232-7389
Diani Kartini https://orcid.org/0000-0001-8660-3088

Abstract

Xeroderma Pigmentosum (XP) is a rare inherited skin malignancy with no causative treatment options. Reporting a 23-year-old woman with *xeroderma pigmentosum* who underwent several surgical tumor removals followed by further five procedures to apply tissue expander, facial resurfacing using full-thickness skin grafts with a donor from abdominal. The next series was tumor resection in the chin, forehead, nasal reconstruction, tumor removal on bilateral third digits, and defect closure. No recurrence after nine years of postoperative monitoring with an aesthetically acceptable result.

Key words: Xeroderma Pigmentosum, Squamous Cell Carcinoma, Basal Cell Carcinoma

Introduction

Xeroderma pigmentosum (XP) is a rare irreversible genetic disorder caused by defective DNA repair from damage attributable to ultraviolet (UV) light. Exposure to sunlight often causes dry skin (xeroderma) and skin color changes (pigmentation) in affected individuals. The genetic abnormality may accumulate and be uncorrected. According to Kleijer et al., the prevalence of XP in American and European countries was around 1: 1,000,000.[1-3] There is no guideline in the treatment of the abnormality due to difficulties in conducting controlled studies with the small number of cases. However, some studies suggest that surgical intervention must always be considered the first-line treatment in a cutaneous tumor and replacing the entire surface of the facial skin with skin that is unexposed to sunlight, such as the skin on the abdomen or buttocks.[4-7]

This article reports a case of XP patient manifested as squamous cell carcinoma (SCC) transitioned to basal cell carcinoma (BCC) managed by tumor excision and facial surface resurfacing with skin grafting with the autologous full-thickness skin graft (FTSG). After nine years postoperatively, no recurrence of skin tumor, graft rejection, or clinical signs of metastasis. This report discussed the contributing factors to the recurrence of this skin malignancy based on studies, showing the beneficial effects of surgeries to prevent tumor recurrence.

Case illustration

A 23-year-old female with xeroderma pigmentosum was admitted on postoperative control following tumor removals and reconstructions. Historically, when she was three months old, she had reddish spots throughout her body. The spots darkened when it exposed directly to sunlight. In 2008, when she was 12 years old, a lump appeared on the nose representing an enlarged nose. Over time, the lesions grew to be more abundant up to 4x14x0.5 cm in the largest– and 1x2x0.5 cm in the smallest size.

On the physical examination, the eyes were shown hyperaemic. Generalized black-red maculopapular lesions were found all over the body, no signs of metastasis or neurological symptoms were found. The working diagnosis was squamous cell carcinoma with suspicion of XP.

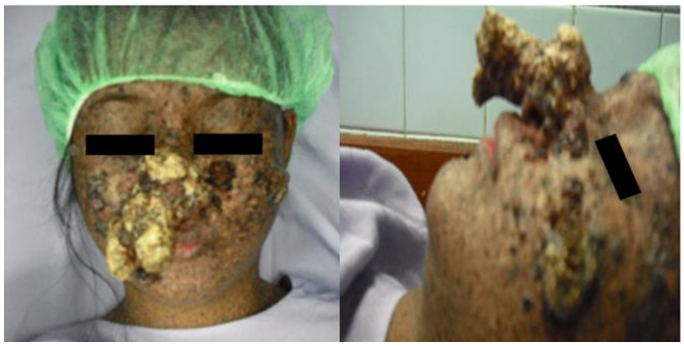

Figure 1. Anterior and lateral view case with skin malignancy before surgery.

Further investigations showed normal laboratory findings. CT scan showed the extension of the left dorsal nasal lesion into the anterior wall of the left maxillary sinus. The histopathology findings revealed disparate results over multiple tests. The tumor on the nose and cheek was surgically resected in 2010. Histopathology showed a well-differentiated, keratinized squamous cell carcinoma with an unclean surgical margin. In 2012, another histopathology finding confirmed XP diagnosis with epithelial cells suggestive for basal cell carcinoma. Another histopathology finding in 2014 confirmed an XP with basal cell carcinoma. Finally, in 2017, histopathology showed a basal cell carcinoma with a tumor-free surgical margin.

She had received six surgical procedures in nine years (2010 to 2019), namely, surgical removal of squamous cell carcinoma in the nose,

cheeks, and both hands, insertion of tissue expander in the abdominal region, forehead, and chin, facial resurfacing with FTSG with the donor in the abdominal, and nasal reconstruction. She did not receive any chemotherapy or radiotherapy afterward. The following procedure was the surgical release of symblepharon of the right eye, and the defect was covered with lip mucosal skin grafts.

Figure 2. Tumor removal on nasal and cheeks (left) and following skin grafting (right).

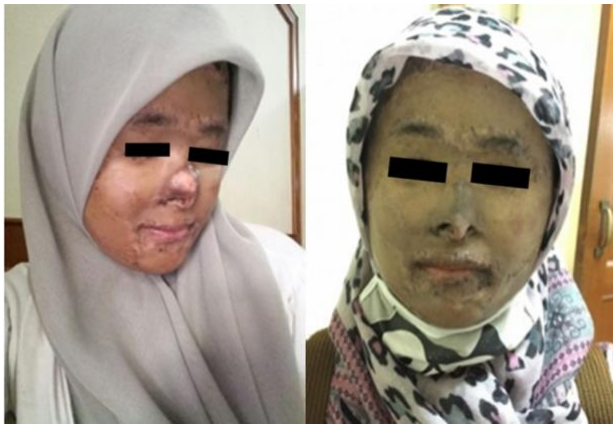

Figure 3. Nine years follow–up showed no recurrence of malignancy.

Discussion

Surgical management remains the first method of treatment in XP manifested with malignancy in the skin. The surgical procedure includes resurfacing with different methods: dermatome shaving followed by dermabrasion and split-thickness skin grafts, partial-thickness skin grafts, full-thickness skin grafts, and sensate-free vascularize fascia–cutaneous radial forearm flaps.[3,8]

They were principally resurfacing to place new tissue or skin strands from regions less exposed to sunlight. The donor skin graft may be autograft or allograft. All studies used skin grafts (autografts) to cover defects after XP. Still, Amin et al. reported using radial forearm free flap (i.e., a sensate-free vascularize fasciocutaneous radial forearm free flap) with a donor from an ABO compatible mother to her five-year-old child with XP.[9] The flap survived in early postoperative days, and the wound heals gradually. But, in the next two weeks, reddish spots appeared on the flap where the biopsy was taken and showed a minimal immunological reaction. Skin rejection was an issue found on the face at the flap site, even with an immunosuppressant. The affected site was retreated surgically using a partial thickness skin graft, and the tumor recurred after ten months.[9]

Some studies showed that basal cell carcinoma in XP was more common at a younger age, and recurrence was more prevalent in regions with more lesions than non-XP.[10] It was contradictory with this reported case. This case earlier was squamous cell carcinoma and changed to basal cell carcinoma within years. She underwent surgery for basal cell carcinoma of the face with a donor autograft from the abdomen using the full-thickness skin graft method and found no recurrence.

Generally, in defects covered using the autograft, the malignancy may be redeveloped, particularly those exposed to sunlight or other rays that may elicit DNA damage on the tumor site. The skin cells in XP are unable to repair damaged NER genes. Thus, allografts from an average skin person will have a lower recurrence than autografts.[10] With the application of the full-thickness skin graft, this report represented a better-healed wound based on skin grafting theory. The full-thickness skin graft may control the development of the tumor, and the skin structure provides more normal skin constituents. In addition, a full-thickness skin graft is more durable and aesthetically beneficial as it has a skin color that matches the slight changes in pigmentation. Thus, the donor area remains well lubricated from its natural sebaceous and sweat glands.

Some reports by Ozmen et al., Agrawal et al., Atabay et al., and Yosipovitch et al., showing no recurrence within an average of two to four years than those reported by Ergűn et al. and Ashall et al. The last reports showed a high recurrence rate despite the use of full-thickness skin graft.[11-16] This prompted other factors to be understood to prevent recurrence in XP. Ergűn et al. showed that donor selection was essential for the outcome. Thus, the donor should be in an area free of pigmentation and less exposed to sunlight.[15] This is crucial because the XP skin exposed to extreme sunlight was more susceptible to damage to genes, providing an unsuitable for a donor purpose. Exposure to light with halogen sources, halides, or fluorescence must also be avoided if an area were addressed to be used as a donor.[9] The donor can be retaken after a complete reepithelialization. Epithelial cells migrate from the remaining hair follicles, sebaceous glands, and sweat glands left in the dermis layer, which generally occurs within seven to ten days; some cases require up to 21 days depending on the patient's age and nutritional status.[17]

Basal cell carcinoma is an aggressive type of cancer with a high recurrence rate, albeit not as aggressive as malignant melanoma. A study by Yosipovitch reported XP manifested with malignancy, namely basal cell carcinoma and malignant melanoma treated with radical tumor excision followed by radiotherapy. No recurrence was reported for up to five years of monitoring. However, other tumors developed from another region that had not been surgically removed during the monitoring period.[14] This is in line with this reported case, in which no recurrence within nine years of postoperative monitoring.

Yosipovitch et al. showed that the autograft used, and defects have the same DNA properties as those of the recipient's body, as previously mentioned. However, those did not regrow into skin malignancy even after sun exposure.[14] This may be due to underlying differences of malignancy in subjects with and without XP. A study showed that XP encountered issues of abnormal cellular-level immunity and delayed hypersensitivity. The study also showed the impaired proliferative response of lymphocytes against mitogens and decreased NK cell activity. Immunohistochemical staining showed complete infiltration of *CD163+ M2* macrophages along with regulatory *Foxp3+* T cells.[18] Nonetheless, there are yet studies to accurately determine lymphocyte profiles in XP. With more studies, the medical world can find an allograft for XP patients to improve their quality of life and survival.

The prognosis includes the association of climate differences in countries with XP cases, unavailability of a simple detection method, no detailed profile of lymphocytes in XP, and the lack of updated surgical techniques and adequacy in excision. To date, full-thickness skin grafts remain the method to treat the skin defect after XP removal.

Conclusions

Even without adjunct therapy, precise surgery yielding surgical free margin could achieve remission in XP. Choice of donor types and usage of FTSG yields favorable results regarding malignancy control.

Disclosure

Authors declare no conflict of interest

Acknowledgment

A sincere appreciation and gratefully acknowledged to the Department of Pathology Anatomy staff for providing and interpreting the samples in diagnosing skin malignancies.

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