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OPEN OLGU SUNUMU / CASE REPORT

Full Facial Resurfacing Followed by Enucleation in a Patient with Xeroderma Pigmentosum: A Case Report

Xeroderma Pigmentosum Tanılı Bir Hastada Tam Yüz Cilt Yenilemesi ve Enükleasyon: Olgu Sunumu

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ÖZET

Kseroderma Pigmentosum, DNA onarım mekanizmalarının bozukluğu ile karakterize nadir görülen otozomal resesif geçişli bir genetik hastalıktır. Bu hastalık, ultraviyole (UV) ışınlarına karşı aşırı hassasiyetle kendini gösterir ve genellikle çocukluk çağında başlar. UV kaynaklı DNA hasarının onarılamaması sonucu, bu bireylerde ciltte erken yaşta malign lezyonlar gelişir. Özellikle güneşe maruz kalan bölgelerde skuamöz hücreli karsinom, bazal hücreli karsinom ve malign melanom gibi cilt kanserleri sıkça görülür. Bu yazıda, 26 yaşında, Orta Doğulu kadın bir hastada ortaya çıkan Kseroderma Pigmentosum olgusu sunulmaktadır. Hasta, yüz bölgesinde tekrarlayan cilt tümörleri ile başvurmuş olup, bu lezyonlar hem estetik açıdan deformitelere hem de genel sağlık açısından riskler oluşturmuştur. Hastanın yaşam kalitesini artırmak amacıyla, kısmi kalınlıkta deri grefti kullanılarak tam yüz yeniden yüzeylendirme (resurfacing) işlemi uygulanmıştır. Uygulanan cerrahi müdahale, yeniden yüzeylendirilen ciltte yeni tümörlerin gelişmesini engellemeyi başarmıştır. Ancak, takip sürecinde hastanın sol göz küresinden dışa doğru büyüyen bir kitle gözlemlenmiştir. Yapılan değerlendirmede, bu kitlenin Kseroderma Pigmentosum hastalarında sıklıkla karşılaşılan agresif ve hızlı ilerleyen cilt tümörlerinin bir sonucu olduğu anlaşılmıştır. Görme fonksiyonlarını tehdit etmesi ve çevre dokulara yayılma riski nedeniyle, enükleasyon (göz küresinin çıkarılması) önerilmiştir. Bu olgu, Kseroderma Pigmentosum hastalarında yüzey yenileme işlemlerinin olumlu etkilerini gösterirken, aynı zamanda tümör gelişiminin agresif seyrine de dikkat çekmektedir. Erken tanı ve düzenli takip tedavi başarısını artıran temel unsurlardır. Bildiğimiz kadarıyla, hem yüz yenileme (resurfacing) hem de enükleasyonun aynı hastada uygulandığı nadir bir vakadır.

Anahtar Kelimeler: Xeroderma Pigmentosum,Enükleasyon,Yeniden yüzeylendirme,Skuamöz Hücreli Karsinom,Malign Melanom

ABSTRACT

Xeroderma Pigmentosum is a rare autosomal recessive genetic disorder that marked by defective DNA repair mechanisms. We present a unique case of Xeroderma Pigmentosum in a 26-year-old Middle Eastern female who exhibited recurrent skin tumors, leading to substantial cosmetic and health-related challenges. The decision to perform a facial resurfacing operation with partial thickness skin graft was made to address the multiple lesions on the face and to improve the patient's quality of life. The procedure was successful in preventing the development of new tumors on the resurfaced skin during the follow-up period. However, one year following the resurfacing operation the subsequent development of a mass protruding from the patient's left globe highlights the aggressive nature of tumors in patients with Xeroderma Pigmentosum. Enucleation was recommended to prevent further complications. To our knowledge, this is a rare case in which both facial resurfacing and enucleation were performed concurrently in a single patient.

Keywords: Xeroderma Pigmentosum, Enucleation, resurfacing, Squamous Cell Carcinoma, Malignant Melanoma,

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INTRODUCTION

Xeroderma Pigmentosum is a rare autosomal recessive genetic disorder that is characterized by defective DNA repair mechanisms (1). The condition was first described in 1874 by dermatologist Moriz Kaposi, who reported four patients presenting with dry, thin, and wrinkled skin, irregular pigmentation, and the development of skin tumors. Over time, further case studies revealed a broader spectrum of symptoms, including progressive neurological degeneration in some patients. The clinical presentation of XP can vary widely—from isolated UV sensitivity and cutaneous lesions to more severe forms that include neurological deficit, dwarfism, gonadal hypoplasia, and intellectual disability (2).

Currently, there is no definitive treatment for Xeroderma Pigmentosum. Medical and surgical approaches aim to alleviate the symptoms of the disease, improve the quality of life, and eventually increase the survival rate of these miserable patients. Multi-disciplinary collaboration with dermatologists, plastic surgeons, neurologists, psychiatrists and ophthalmologists is

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essential to provide comprehensive care for these patients and to prevent the development of potentially life-threatening complications.

CASE

This case report is to describe a rare case of Xeroderma Pigmentosum in 26-year-old female patient presenting with recurrent skin tumors to the Department of Plastic Reconstructive and Aesthetic Surgery.

The patient developed her initial lesion at the age of seven, when she got diagnosed after being on a summer vacation to the beach. Afterwards the patient developed several lesions which were excised locally. Over the years, and with the wars in the middle eastern region, the patient had limited access to sun

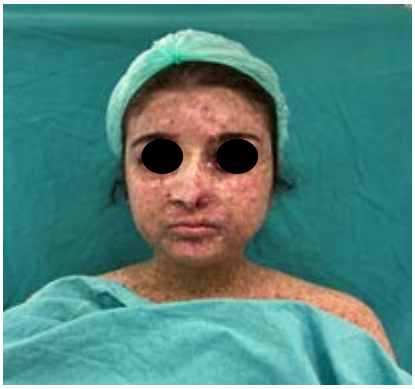

Figure 1. Shows the preoperative appearance of our patient

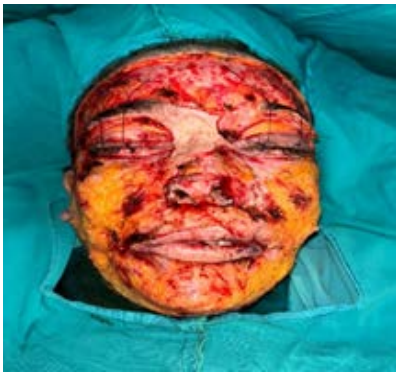

Figure 2. Shows the intraoperative appearance after skin excision

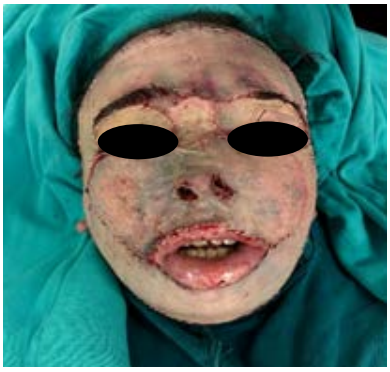

Figure 3. Shows the appearance after the graft was applied to the recipient area

Figure 4. Shows the excised skin

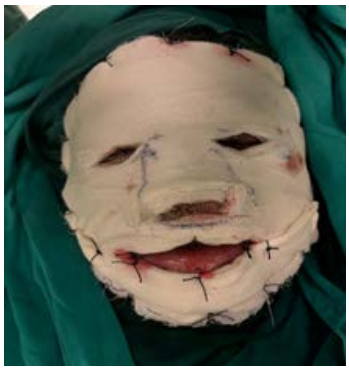

Figure 5. Shows the tie over dressing

Figure 6. Shows the preoperative appearance of the globe Squamous Cell Carcinoma

Figure 7. Shows the intraoperative appearance during the enucleation procedure

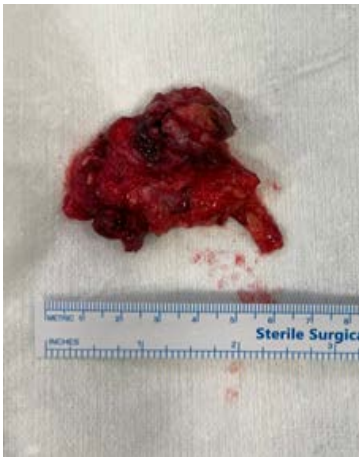

Figure 8. Shows the enucleated globe

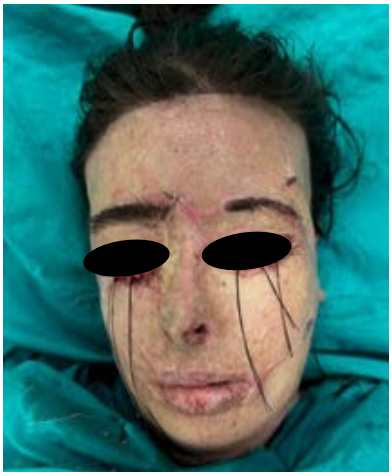

Figure 9. Shows the postoperative appearance after the enucleation procedure

Figure 10

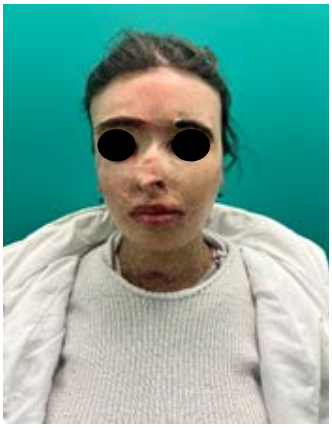

Figure 11

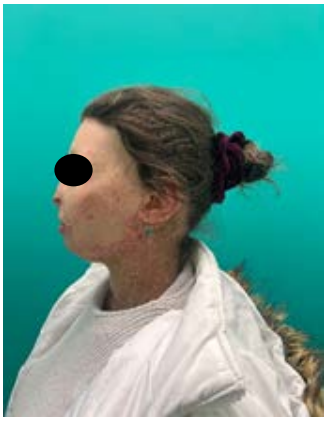

Figure 12

Figure 10,11,12. Shows the postoperative first month appearance after the enucleation procedure

Figure 13

Figure 14

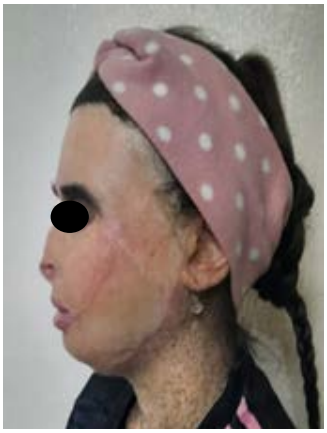

Figure 15

Figure 13,14,15. Show the postoperative second-year appearance after the resurfacing procedure

protection, the patient moved as a refugee to Kahramanmaras, Turkey, where she was followed up in our institution, later with the occurrence of the devastating 2023 earthquake in southeastern Turkey, the patient's condition even worsened.

The patient initially presented to our hospital eight years ago with a lesion on her dorsal nasal area which was locally excised by our team and the defect was closed by a full -thickness skin graft from her inguinal region. Afterward the patient presented 17 times for local excision of various tumors including Basal Cell Carcinoma, Squamous Cell Carcinoma, Malignant Melanoma, Keratoacanthoma,and Collagenoma, which were excised locally, and the defects were closed primarily. The patient's recurrent skin tumors posed significant cosmetic and health concerns. Due to the development of these recurrent lesions especially on the face of the patient except on the grafted area we applied 8 years ago, we decided to perform a facial resurfacing operation with split-thickness skin graft from the patient's abdominal area. As the patient dressed decently, her abdominal area was thought to be UV- protected Fig.1 shows the preoperative appearance of our patient. The patient and her family were notified about the advantages and possible complications of this procedure, and they accepted the procedure. Our operation was done with respect to the aesthetic facial subunits preserving the periorbital skin, the previously grafted area and the lips of the patient fig.2,3. The procedure was done, and the excised full-face skin was sent for pathological examination fig. 4. Postoperatively the patient was followed-up with tie-over dressing using paraffine mesh Fig.5, and after wounds healed uneventfully, the patient was advised to perform massage using silicone-based anti-scar jells to improve the cosmetic outcome fig.6 shows the postoperative 1- year result.

After performing the procedure, the patient did not develop any malignant tumors in her resurfaced skin. However, one year later she presented with a mass protruding from her left globe figure 6. Enucleation procedure was recommended to the patient, and she accepted the procedure with no hesitation. The globe along with the lacrimal gland were sent to pathological examination fig.8 which revealed minimal invasive Squamous Cell Carcinoma, and the lacrimal gland was tumor free. The postoperative period was uneventful, and the patient got discharged on the third postoperative day Fig.9. Fig.10 ,11 and 12 show the postoperative first month appearance of the patient. Fig,13,14 and 15 show the postoperative second year appearance after the initial resurfacing procedure.

DISCUSSION

Xeroderma Pigmentosum (XP) is a rare autosomal recessive genetic disorder caused by defects in the DNA nucleotide excision repair pathway, resulting in an impaired ability to repair UV-induced DNA damage. Clinical manifestations of Xeroderma Pigmentosum (XP) generally appear in early childhood, with key signs including freckling before the age of two and severe sunburn after even minimal exposure to sunlight. Individuals with XP face a markedly elevated risk of developing skin cancers at an unusually young age. Non-melanoma skin cancers often arise around the age of nine, while melanoma tends to appear by the early twenties. In our case the patient developed her first malignant melanoma at the age of 22 which was invasive type superficial spreading subtype of on the dorsal aspect of her right hand. In addition, individuals with Xeroderma Pigmentosum often experience premature skin aging, characterized by symptoms such as thinning, dryness, telangiectasia, and uneven pigmentation (2). In addition to the autosomal recessive version of XP a dominant version has also been reported in a Scottish female with a milder clinical course (3).

Currently there is no definitive treatment for Xeroderma Pigmentosum. Genetic counseling and prenatal diagnosis are essential for preventing the occurrence of the disease. In our case the patient was the only affected member of her family, and she did not have a family history of such condition. Several treatments have demonstrated a reduction in the severity of the course of the disease including less invasive treatments as , topical 5-fluorouracil, oral retinoids, chemical peeling, dermabrasion in addition to surgical treatment by simple excision, sub-total facial excision and resurfacing by composite tissue allotransplantation, full thickness and split thickness skin grafts(3,4,5,6) . Grafts are typically harvested from sun-protected areas of the body, such as the abdomen, thighs, and buttocks, which are presumed to be the least exposed to sunlight. Full-thickness skin grafts are considered cosmetically superior to split-thickness grafts due to their lower tendency for secondary contraction, as well as better-preserved pigmentation, sebaceous, and sweat glands in the recipient area. However, split-thickness grafts offer an advantage in terms of allowing easier surveillance for the recurrence of skin cancer in the grafted site. (7).

Ocular manifestations are present in approximately 40–100% of individuals with Xeroderma Pigmentosum (XP), ranging from benign degenerative changes to more severe ocular and periocular malignancies. The most observed malignant tumor affecting the ocular surface is ocular surface squamous neoplasia (OSSN). Risk factors for the development of OSSN include prolonged exposure to ultraviolet (UV) radiation, immunosuppression (such as in HIV infection or chronic corticosteroid use), XP, infection with Human Papillomavirus (HPV), heavy tobacco use, male gender, and advanced age. Diagnosis is usually clinically with definitive diagnosis made with a biopsy (8,9,10). In our case the patient carried two of the risk factors above where in addition to being a XP patient, she had a history of chronic use of ophthalmic steroids drops.

Conclusion

Xeroderma Pigmentosum (XP) is a rare autosomal recessive disorder characterized by defective DNA repair mechanisms, particularly in response to ultraviolet (UV) light-induced damage. Clinically, the condition manifests as severe hypersensitivity to UV radiation, with involvement of the ocular system and, in some cases, progressive neurological deterioration. Given the absence of a definitive cure, management is predominantly focused on conservative strategies, with strict UV protection being the cornerstone of

care to minimize the risk of skin and ocular complications (11). The case presented underscores the profound impact of Xeroderma Pigmentosum on both the physical and psychological well-being of patients, especially in the context of recurrent and potentially life-threatening skin malignancies and especially taking into consideration the gender of our patient. Despite the challenges posed by the disease, early intervention, rigorous UV protection, and advanced techniques such as skin resurfacing surgery can significantly improve both cosmetic outcomes and the patient's overall quality of life. The successful management of this patient highlights the necessity of a multidisciplinary approach, involving dermatological screening, surgical intervention, and psychological support, to address both the medical and emotional needs of individuals with XP. Continuous surveillance and preventive care are critical in reducing the risks associated with this disorder and improving patient outcomes.

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Patient consent permission: *We have a signed copy of the form of the patient consent permission in this case report.*

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