Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Giant Squamous Cell Carcinoma in Xeroderma Pigmentosum: A Case Report and Literature Review

Tariq Igarramen*, Oumayma Bounid, Mouna Darfaoui, Issam Lalya, Abdelhamid El Omrani, Mouna Khouchani

Department of Radiation Oncology, Mohammed VI University Hospital, Marrakech, Morocco

DOI: 10.36347/sjmcr.2021.v09i08.001 | **Received:** 29.06.2021 | **Accepted:** 01.08.2021 | **Published:** 03.08.2021

***Corresponding author:** Tariq Igarramen

Abstract **Case Report**

Xeroderma pigmentosum is a rare autosomal recessive genodermatosis characterized by photosensitivity and the development of cutaneous and internal malignancies at an early age. The clinical manifestations basic defect is a nucleotide excision repair defect, leading to deficient repair of DNA damaged by ultraviolet radiation. These patients exhibit increased sensitivity to ionizing radiation Patients with xeroderma pigmentosum who are younger than 20 years old have a significantly higher than a 1000-fold increased risk of developing skin cancer. Early diagnosis of these tumors is crucial because they are fast-growing, early metastasizing, and lead to death. However, early detection and treatment of cutaneous malignancies will decrease morbidity and mortality. Genetic counselling remains the most crucial measure for preventing xeroderma pigmentosum. We report a case of xeroderma pigmentosum in a 17-year-old girl presenting with a giant malignant tumor on her face, emphasizing the importance of early diagnosis and management.

Keywords: Xeroderma Pigmentosum, Squamous Cell Carcinoma, Oncology.

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disease, characterized by a deficiency in DNA repair mechanism through the nucleotide excision repair (NER) pathway, which leads to sensitivity to ultraviolet (UV) radiation, thereby promoting the appearance of cutaneous tumors, such as squamous cell carcinoma (SCC), basal cell carcinoma (BCC), and cutaneous melanoma (CM) [1, 2].

XP is clinically characterized by extreme skin photosensitivity, pigmentary anomalies of skin exposed to sunlight, and ophthalmic and neurological damage [3, 4]. The prevalence of XP is relatively high in the Middle East and Japan [3]. A few cases have also been previously reported in Africa [5, 6]. In this region, the more intense sun exposure predisposes people to early onset of squamous cell carcinoma despite pigmentosa protection [3]. The occurrence of these cancers is related to a defect in DNA repair. We report a case of xeroderma pigmentosum in a 17-year-old girl presenting with a giant malignant tumor on her face, emphasizing the importance of early diagnosis and management.

PATIENT AND OBSERVATION

A 17 years old girl presented to our hospital with a one-year history of rapidly growing masse in her lateral hemiface. She was born from first-degree blood relatives, and she and her brother were followed up for four years for XP.

A dermatological examination revealed an ulcerating mass in the right hemiface, measuring about 10×15 cm, with irregular edges. The full-body skin was unnaturally dry and rough. Hyperpigmented hyperkeratotic changes were observed over some lesions (figure 1).

Computed tomography (figure 2) demonstrated

On the cervicofacial level

Ulcerated and budding tumor of the right hemiface, with a maximum thickness of 50 mm and extended over 120 mm, hetero-dense enhancing in a heterogeneous way containing very hypodense zones.

Topographically

- It invades the orbit and the lacrimal gland and encompasses the eyeball on the right side.

- It infiltrates the right retro-maxillary-zygomatic space. It presents an intimate contact with the nose root and the maxillary bone with an irregular cortical bone.
- It infiltrates the face soft tissues and extends to the right atrial region
- Respect the parotid and sub mandibular gland It
- Multiple and bilateral cervical lymph node formations not exceeding 9 mm for the larger ones.
- No other localization on the thoracic and the abdominal level

The tumor was not resectable, and the decision was to start Metotrexat due to the altered general states of the patient.

DISCUSSION

XP affects both males and equally and is commonly symptomatic since childhood [7]. In Western Europe, the United States, and Japan, the incidence of XP is 1/500 000, 1/250 000, and 1/22,000, respectively [8]. Moreover, having a greater incidence in populations where consanguinity is prevalent [9].

The clinical symptoms of XP vary considerably, and the cumulative UVR exposure determines disease severity and age of onset [10].

Defects in nucleotide excision repair cause hyperpigmentation, hypopigmentation, lentigos, telangiectasias, actinic keratoses, and atrophy due to premature sunlight-induced damage [11]. Cutaneous signs generally appear before the age of two, and the first skin malignant neoplasia usually appears in the first decade of life [11]. XP increases the frequency of skin cancer in >10,000-fold for non-melanoma skin cancer (NMSC) and >2000- fold increased risk of melanoma in patients under 20 years of age compared to the general U.S. population [11-13]. In the XP patients, the median age at diagnosis of first NMSC was nine years (range 1–32 years) and melanoma was 22 years (range 2–47 years) [9], showing that XP patients had a 58 year reduction in age at first NMSC and a 33 year reduction in age at first melanoma [9].

Eyelid and conjunctival cancers have been reported [9-11], like the interpalpebral squamous cell carcinoma. Limbal tumor includes squamous cell carcinomas, pterygia, and malignant melanoma. Melanoma may rarely affect the iris. Orbital tumors include squamous cell carcinomas, basal cell carcinomas, and melanomas [11, 14].

The avoidance of UVR exposure from sunlight is recommended for preventing these neoplasms in patients with XP through UVR-protective clothing, hats, gloves, and sunglasses, the application of high-factor sunscreen, and UVR-blocking window films at home and in cars.

CONCLUSION

An interprofessional team is necessary for the management of patients with xeroderma pigmentosum. Because of their considerably higher risk of developing malignant tumors, several physicians are required for the diagnosis and management of these tumors in order to offer the best clinical result and quality of life for each patient.

Fig-1: Ulcerating mass in the right hemiface, measuring about 10×15 cm

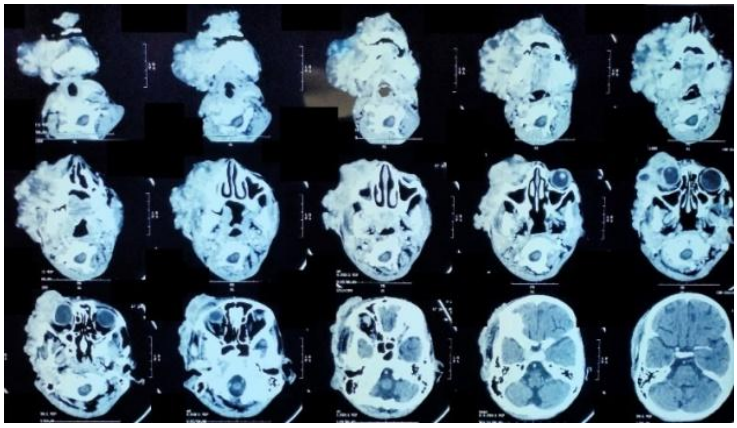

Fig-2: CT scan

REFERENCE

1. Miraglia, E. (2013). Xeroderma Pigmentosum: The Star&#xfffd;s Sons. Case Report and Review of the Literature. P.& R. 2013. https://doi.org/10.11138/PR/2013.2.3.111.
2. Soltys, D. T. (2010). Análise da natureza genotípica de pacientes Xeroderma Pigmentosum brasileiros (Doctoral dissertation, Universidade de São Paulo).
3. Kraemer, K. H., Lee, M. M., & Scotto, J. (1987). Xeroderma pigmentosum: cutaneous, ocular, and neurologic abnormalities in 830 published cases. Archives of dermatology, 123(2), 241-250.
4. Pandey, A., Kuldeep, K., Koul, A., Tyagi, M., Singh, P., Sharma, P., & Dimri, D. (2013). Xeroderma pigmentosa with ocular association: case report. Case Rep Clin Med, 2, 466-469.
5. Dieng, M. T., Niang, S. O., Dangou, J. M., & Ndiaye, B. (2001). Xeroderma pigmentosum: à propos de 6 cas observés à Dakar. Bulletin du cancer, 88(2), 199-202.
6. Chidzonga, M. M., Mahomva, L., Makunike-Mutasa, R., & Masanganise, R. (2009). Xeroderma pigmentosum: a retrospective case series in Zimbabwe. Journal of oral and maxillofacial surgery, 67(1), 22-31.
7. Brooks, B. P., Thompson, A. H., Bishop, R. J., Clayton, J. A., Chan, C. C., Tsilou, E. T., ... & Kraemer, K. H. (2013). Ocular manifestations of xeroderma pigmentosum: long-term follow-up highlights the role of DNA repair in protection from sun damage. Ophthalmology, 120(7), 1324-1336.
8. Lim, R., Sethi, M., & Morley, A. M. (2017). Ophthalmic manifestations of xeroderma pigmentosum: a perspective from the United Kingdom. Ophthalmology, 124(11), 1652-1661.
9. Schubert, S., Lehmann, J., Kalfon, L., Slor, H., Falik-Zaccai, T. C., & Emmert, S. (2014). Clinical utility gene card for: Xeroderma pigmentosum. European Journal of Human Genetics, 22(7), 953-953.
10. DiGiovanna, J. J., & Kraemer, K. H. (2012). Shining a light on xeroderma pigmentosum. Journal of investigative dermatology, 132(3), 785-796.
11. Ramkumar, H. L., Brooks, B. P., Cao, X., Tamura, D., DiGiovanna, J. J., Kraemer, K. H., & Chan, C. C. (2011). Ophthalmic manifestations and histopathology of xeroderma pigmentosum: two clinicopathological cases and a review of the literature. Survey of ophthalmology, 56(4), 348-361.
12. Bradford, P. T., Goldstein, A. M., Tamura, D., Khan, S. G., Ueda, T., Boyle, J., ... & Kraemer, K. H. (2011). Cancer and neurologic degeneration in xeroderma pigmentosum: long term follow-up characterises the role of DNA repair. Journal of medical genetics, 48(3), 168-176.
13. Cleaver, J. E., Lam, E. T., & Revet, I. (2009). Disorders of nucleotide excision repair: the genetic and molecular basis of heterogeneity. Nature Reviews Genetics, 10(11), 756-768.
14. Rizvi, S. A., Amitava, A. K., Mehdi, G., Sharma, R., & Alam, M. S. (2008). Orbital amelanotic melanoma in xeroderma pigmentosum: A rare association. Indian journal of ophthalmology, 56(5), 421.