IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 23, Issue 6 Ser. 6 (June. 2024), PP 24-27
www.iosrjournals.org

Ocular Damage In Xeroderma Pigmentosum: A 40-Eye Study In Morocco

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Abstract
Xeroderma pigmentosum (XP) is a rare genetic disease characterized by hypersensitivity to ultraviolet (UV) radiation due to a defect in DNA repair. While the increased risk of skin cancer is well documented, ocular manifestations of this disease are less common but can have a significant impact on the quality of life of patients.
This article presents retrospective study of 40 eyes of Moroccan patients with XP and significant ocular involvement, highlighting the importance of awareness, prevention, early detection, and interdisciplinary collaboration.
Keywords: *xeroderma, pigmentusum, ocular , eyelid, conjonctival*

Date of Submission: 07-06-2024 Date of Acceptance: 17-06-2024

I. Introduction

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterized by hypersensitivity to ultraviolet (UV) radiation due to a defect in DNA repair. While the increased risk of skin cancer is well documented, ocular manifestations of this disease are less common but can have a significant impact on the quality of life of patients.
In this article, we present a study of 40 Moroccan eyes with XP and significant ocular involvement.

II. Materials and Methods:

We report a retrospective study of 40 eyes collected between 2019 and 2023. Our patients presented with eyelid neoplasms with or without conjunctival involvement. The treatment consisted of oncological tumor excision supported by data from histopathological examinations.

III. Results:

The average age of the patients was 13 years (ranging from 4 to 23 years), with 44% of the patients being female and 56% male, with a sex ratio of 1.22. Most of the cases came from a low socioeconomic background. Consanguinity was found in 75% of the cases, with first-degree consanguinity in 15 cases, second-degree in 10 cases, and third-degree in 5 cases. This was observed in 30% of our patients. In our study, most of our patients were exposed to sunlight.

60% of the patients had ocular and periocular malignant tumors: isolated eyelid tumors (15 cases), conjunctival tumors (17 cases), or a combination of both (15 cases). The involvement was bilateral in 70% of the cases. The histological type varied, with squamous cell carcinoma being the most common (28 cases), followed by basal cell carcinoma (2 cases), squamous cell carcinoma (1 case), and carcinoma in situ (2 cases). Other benign lesions included symblepharon in one case, conjunctival hemangioma in one case, and retractile ectropion or entropion in 2 cases. 10 eyes underwent excision treatment due to local-regional invasion.

IV. Discussin

25 patients underwent biopsy with complete excision treatment using mitomycin C for conjunctival involvement. The postoperative course was marked by the stabilization of lesions in the majority of cases.

XP is much more common in the Middle East due to consanguineous marriages [1]. Consanguinity was found in 75% of cases, which is common in North Africa.

Visual system involvement occurs in 40% of XP patients [2], with severity correlated to the extent of cutaneous involvement and based on the same pathological mechanisms. Ocular involvement is therefore bilateral and limited to the anterior segment: conjunctiva, eyelids, and cornea, which are the tissues exposed to UV [3, 4].

The severity of involvement varies, with lesions often being bilateral and localized only in the anterior segment. This confirms literature findings and highlights the importance of ocular involvement as a characteristic of XP [3].

Symptoms are progressive, and photophobia is the most consistent and earliest sign, even before the appearance of cutaneous manifestations. This allows for early diagnosis in at-risk families. The mechanism of photophobia is unknown, but it is often intense and requires a specific response: keeping the head down, partially open and teary eyes while seeking darkness. This photophobia tends to decrease as corneal opacity develops. All patients in our series reported photophobia, but it was not the only diagnostic clue before the onset of cutaneous involvement [3].

The eyelids are primarily affected and exhibit all the cutaneous lesions of XP, including signs of blepharitis, dyschromic changes, atrophies leading to ectropion or retractile entropion. As a result, the eyelids progressively lose their protective function, worsening the ocular prognosis. They can also be the site of benign tumors (papillomas) and especially cutaneous cancers such as epitheliomas or melanomas [4, 5].

In our study, the eyelids were also predominantly affected compared to the rest of the anterior segment. This predominance of eyelid involvement can be explained by their greater exposure to UV rays

The conjunctiva is often affected in cases of squamous cell carcinoma, although rare cases of malignant melanoma have been reported [6]. Basal cell carcinomas have also been observed [5]. These tumors are generally localized around the eyes, but can reach the cornea or even the orbit, presenting a vital (metastasis) and functional risk due to frequent recurrence and extension [3, 4]. Conjunctival melanomas are rare [3, 4].

In most Maghrebian series [7, 8], ocular tumors were observed in 25% of cases, while in Kraemer's series the figure was 11% [9].

Corneal lesions may manifest as keratitis, ulceration, opacity, nodular dystrophy, edema or neovascularization. In our study, we found that 44% of our patients (4 cases) had corneal lesions similar to those described in the literature [4, 10, 11].

Iris involvement is rare in XP, but can manifest as atrophy, altered pigmentation or melanoma [5].

Ocular manifestations can appear as early as the first decade of life and are normal in the general population, where age and prolonged exposure to the sun are the main risk factors. XP therefore accelerates sun-induced ocular aging.

This study underlines the importance of regular ophthalmological surveillance in XP patients, in order to detect ocular damage at an early stage. Symptoms such as photophobia, neovessels, and growths can be early signs of malignancy.

Prevention plays an essential role. XP patients need to be rigorously educated about photoprotection, including wearing sunglasses and regularly applying sunscreen to reduce UV exposure.

A multidisciplinary approach is essential. Ophthalmologists must collaborate with oncologists to ensure optimal management of ocular involvement in XP (12). Treatment options, such as surgery, must be carefully discussed.

V. Conclusion

XP cases with ocular involvement require special attention. This study illustrates the clinical variability of this rare disease, and highlights the importance of awareness, prevention, early detection and interdisciplinary collaboration. It discusses in greater detail the clinical aspects, treatment options, future prospects and clinical implications of these cases.

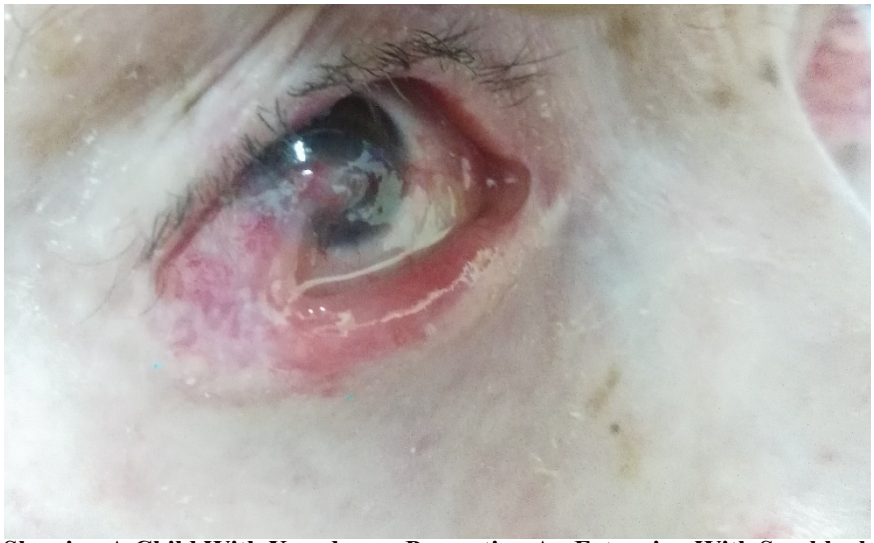

Figure 1 : Showing A Child With Xeroderma Presenting An Ectropion With Symblepharon And Pterygion

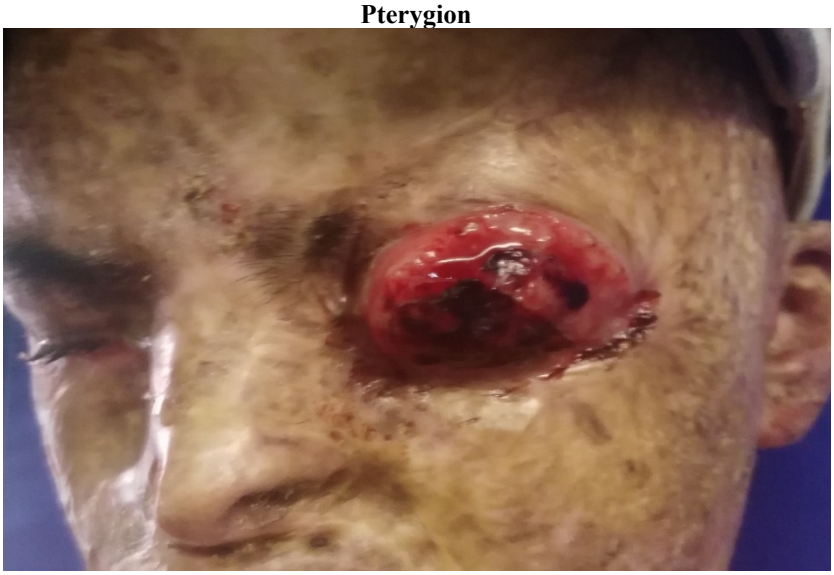

Figure 2 : Photo Of A Child Treated For Xeroderma With Basal Cell Carcinoma Invasive

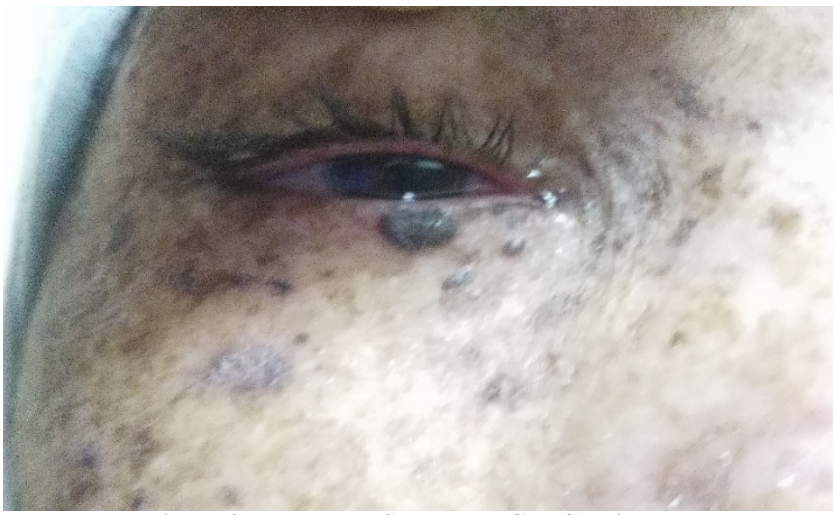

Figure 3 : Palpebral Squamous Cell Carcinoma

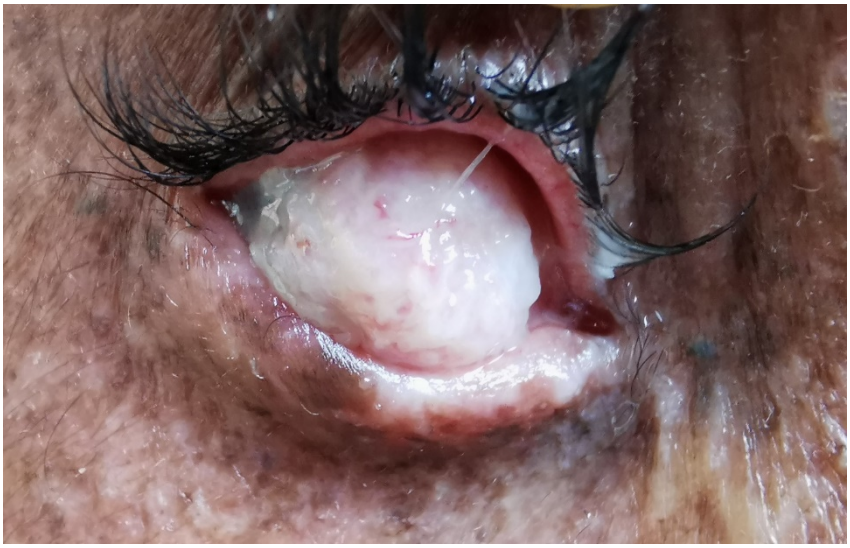

Figure 4 : Conjunctival Squamous Cell Carcinoma Invading The Ocular Surface

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