IJCRT.ORG ISSN : 2320-2882

INTERNATIONAL JOURNAL OF CREATIVE RESEARCH THOUGHTS (IJCRT)

An International Open Access, Peer-reviewed, Refereed Journal

Inflammatory conjunctival tumor in a child with xeroderma pigmentosum: case rapport.

Authors: El marzouqi Batal 1, Amhoud Karim 2, Bouzoubaa Tarik3, Abdallah Elhassan 4, Berraho Amina 5.

Ophthalmology service B, hospital specialties Rabat (CHU Ibn Sina), Mohammed V University, Morocco.

Abstract:

We report the case of a six-year-old child suffering from xeroderma pigmentosum, who presents with swelling of the conjunctiva of the eye. Excisional surgery was performed without any additional treatment. The histological study confirms the inflammatory origin of the tumor.

Xeroderma pigmentosum is a rare photo-dermatosis of genetic origin (autosomal recessive). It is characterized by a defect in the DNA repair system related to extreme sensitivity to ultraviolet (UV) rays. The severity of this disease is linked to the high risk of cancers of the ocular surface and the skin.

Keywords: xeroderma, pigmentosum, tumor, conjunctiva, resection, anatomopathlogy.

Introduction :

Xeroderma pigmentosum (XP) is a rare genetic disorder associated with faulty DNA repair. It is associated with various malignant tumors of the skin and ocular surface such as basal cell carcinoma, melanoma and squamous cell carcinoma. Patients with XP should be assessed regularly to screen for and manage this wide range of malignant tumors.

Eye involvement is common in Xeroderma Pigmentosum, which can affect the eyelids, conjunctiva, cornea or even the eyeball. Basal cellular epitheliomas, but especially spino-cellular epitheliomas, play a significant role in these attacks. Their treatment is based on prevention and surgery. We report the case of a child with XP presenting with a corneo-conjunctival swelling first suggesting a squamous cell carcinoma.

Observation :

We report the case of a six-year-old male child, diagnosed with XP at the age of 6, who consults the ophthalmology department for tearing, photophobia and who presents with the appearance of a mass in the right eye. evolving for 20 days (figure 1,2). According to interrogation, parents have seen multiple discreet dark pigmented lesions on her forehead since the age of 2. Similar lesions gradually appeared on his face, neck, upper and lower limbs. From early childhood, the child has a high sensitivity to bright light and chronic photophobia and tearing. There was no history of inbreeding in the family and no history of similar skin disease in the family. However, he did not seek medical advice until he was 6 years old. A dermato-pediatric opinion was requested.

Ophthalmologic examination reveals a fleshy, budding inferior temporal conjunctival-limbic lesion of the right eye (Figure 3). Faced with the suspicion of the carcinomatous origin of the lesion, an excisional biopsy with pathological analysis was performed, showing that a hyperplastic fleshy bud was found. (Figure 4, 5)

Discussion:

XP was first described in 1874 by Hebra and Kaposi. [1] In 1882, Kaposi coined the term "XP" for the condition referring to his characteristic dry pigmented skin.

XP is inherited as an autosomal recessive disorder. [2] XP is seen during infancy, particularly around 2 years of age with equal sexual incidence. It is a DNA repair disorder in which the ability to repair damage caused by UV light is deficient. If the tumor suppressor genes are affected, the result is fatal cancer such as malignant melanoma, basal cell carcinoma, and squamous cell carcinoma, which are the common causes of death in victims of XP. The average age for skin cancer is 8 years, but actinic damage occurs between 1 and 2 years. The clinical course of the disease can be divided into three stages.

Usually, the first stage appears after the age of 6 months. This stage is characterized by diffuse erythema, desquamation and areas resembling freckles, increased pigmentation in areas exposed to the sun, with the initial involvement of the face. As the disease progresses, skin changes appear on the lower legs, neck, and arms. Although these characteristics tend to diminish during the winter months with a decrease in sun exposure at the initial stage of the disease, they become permanent over time. The second stage is characterized by poikiloderma, which consists of skin atrophy, telangiectasias, and mottled hyper and hypo-pigmentation. The third stage is heralded by the appearance of numerous malignant tumors. [5]

Eye involvement is evident in at least 40% of XP patients, blepharospasm and photophobia are common symptoms. Eyelid skin changes reflect local skin changes including erythema, pigmentation, atrophy, and malignant changes. [5] Telangiectasias, loss of eyelashes and chronic blepharitis are also observed. [5] Atrophic scarred skin can cause ectropion and symblepharon. Lower eyelid loss can lead to exposure keratitis, edema, and even corneal ulceration and perforation. [6] Corneal opacification, neovascularization, pterygium, and band keratopathy are common. Conjunctival involvement usually includes conjunctivitis, pinguecula, symblepharon, melanosis, and tumors developing from the interpalpebral area of the limbus. [5] Limbal tumors are common and squamous cell carcinomas, malignant melanomas, and limbal stem cell deficiency have been reported. The iris can be affected by iritis, stromal atrophy, pigment abnormalities, and rarely malignant melanoma. [7]

Clinical management of XP includes avoidance of sunlight, minimization of exposure to UV and cigarette smoke, early excision of skin lesions, and genetic counseling. [8] Ophthalmic management includes UV absorbing sunglasses with side shields, artificial tears, intermittent topical steroids, monitoring for ocular neoplasms and management of complications. Cancers of the eyelids and conjunctiva are the most frequently reported. Malignant conjunctival tumors that can be excised should be removed and treated with adjuvant cryotherapy / irradiation / topical chemotherapy. [9] Some malignant limbal tumors can be removed by iridocyclectomy, while others may require enucleation. [8] Corneal tumors have been managed with keratoplasty and topical chemotherapy. [8] Choroidal melanoma is commonly managed with plaque radiotherapy, but this has not been specifically studied in XP patients. [8] If a tumor involves the orbit, imaging is necessary and surgical excision with supplemental radiation may be therapeutic. [7] Large or invasive eye or orbital tumors may require enucleation and / or orbital exenteration.

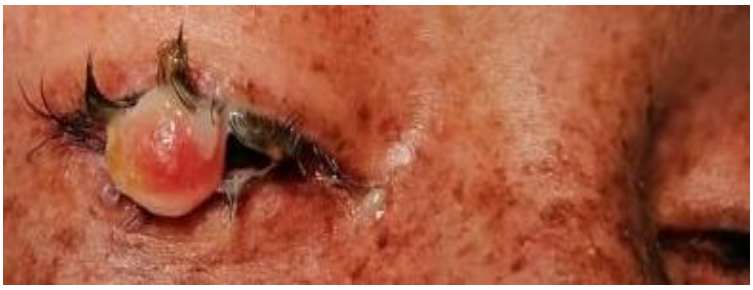
The prognosis for this disease is poor with less than 40% of patients surviving beyond the age of 20. [12] Malignant skin neoplasms are observed in 60% of cases and squamous cell carcinoma in 20% of XP patients. [6] In our case, we first suggested a squamous cell carcinoma, but the pathological study returned in favor of a hyperplastic fleshy bud.

Conclusion:

Xeroderma Pigmentosa, although rare, can present with ophthalmic involvement ranging from benign conjunctival hyperplasia to carcinomatous neoplasia of the cornea, limbus or conjunctiva, for which total excision of the mass with cryotherapy gives good results. . The role of the

ophthalmologist in the early detection and excision of suspected lesions, advice and rapid referral to the dermatologist and oncologist are essential in the management of these patients.

FIGURE :



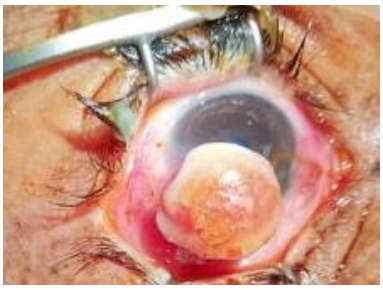


Figure 1- figure 2 : budding conjunctival tumor of the right eye

Figure 3: mass with limbic starting point with narrow base.

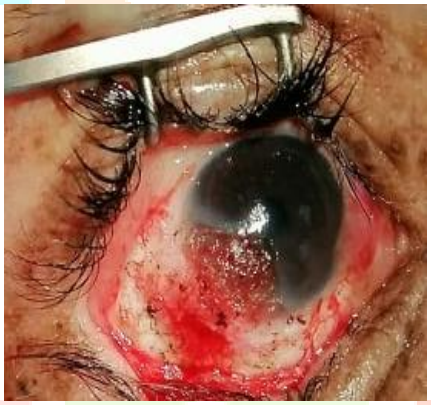


Figure 4: image after resection of the mass.

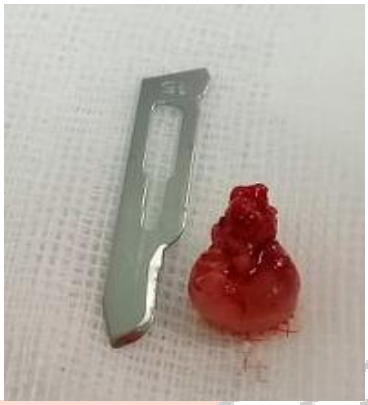


Figure 5: piece sent for anatomopathological examination.

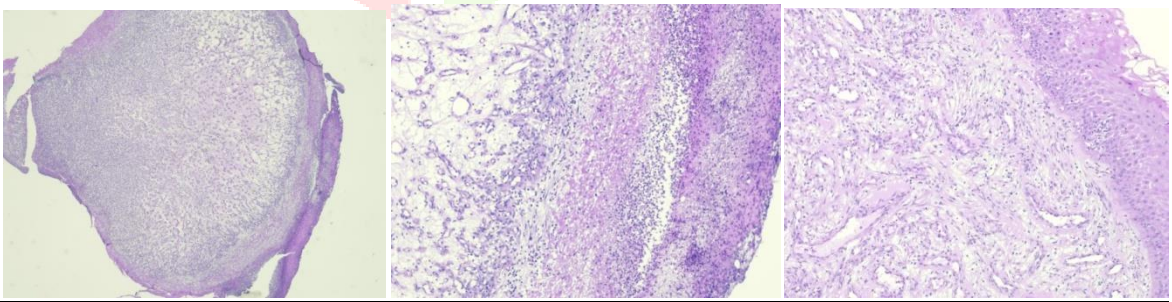


Figure 6: Three anatomy-pathological images of the surgical specimen, increased magnification from left to right, in favor of a hyperplastic inflammatory bud without signs of malignancy.

References:

[1] . Hebra F, Kaposi M. Diseases of the skin including exanthemata. New Sydenham Soc 1874;61:252-8.

[2] . Elder D. System of Ophthalmology. Vol. VIII. London: Henry Kimpton; 1977. p. 551-6

[3] . Kraemer KH. Xeroderma Pigmentosum. In: Pagon RA, Bird TC, Dolan CR, Stephans K, editors. Seattle, WA: Universoity of Washington, Seattle. Gene Reviews; 2008

[4]. Kraemer KH, Lee MM, Scotto J. Xeroderma pigmentosum. Cutaneous, ocular, and neurologic abnormalities in 830 published cases. Arch Dermatol 1987;123:241-50

[5]. Kleijer WJ, Laugel V, Berneburg M, Nardo T, Fawcett H, Gratchev A, et al. Incidence of DNA repair deficiency disorders in western Europe: Xeroderma pigmentosum, Cockayne syndrome and trichothiodystrophy. DNA Repair (Amst) 2008;7:744-50

[6]. Blanksma LJ, Donders PC, van Voorst Vader PC. Xeroderma pigmentosum and keratoconus. Doc Ophthalmol 1986;64:97-103.

[7]. Johnson MW, Skuta GL, Kincaid MC, Nelson CC, Wolter JR. Malignant melanoma of the iris in xeroderma pigmentosum. Arch Ophthalmol 1989;107:402-7

[8]. Kitagawa KO, Inoue M. Choroidal malignant melanoma occurring in a patient with xeroderma pigmentosum. Fol Ophthalmol Jpn 1981;32:657-63

[9]. Kraemer KH, DiGiovanna JJ, Moshell AN, Tarone RE, Peck GL. Prevention of skin cancer in xeroderma pigmentosum with the use of oral isotretinoin. N Engl J Med 1988;318:1633-7.