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Case Report

Xeroderma pigmentosum: a case series with ocular involvement

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ABSTRACT

Xeroderma pigmentosum (XP) is a rare, autosomal recessive disease caused by a defect in DNA repair. Patients with xeroderma pigmentosum often have cutaneous and ocular photosensitivity, freckle-like skin pigmentation, multiple skin and eye cancers, and, in some patients, progressive neurodegeneration. Xeroderma pigmentosum predominantly affects the UV exposed ocular surface, resulting in eyelid atrophy and cancers, corneal dryness, exposure keratopathy, and conjunctival tumors. Hereby, we report four cases of XP with ocular pathology. First case had ectropion, corneal abrasion, keratomalacia, and necrotic ulcer in periorbital area and second case had corneal opacity, conjuctival erythema and photophobia. The other two cases were siblings of second patient who also had photophobia. These cases illustrate the role of DNA repair in protection of the eyes from UV damage.

Keywords: Xeroderma pigmentosum, Opthalmohelioses, Malignancy

INTRODUCTION

Xeroderma pigmentosum (XP), first described by Kaposi in 1870 is a rare, autosomal recessive disease caused by defective DNA repair. The condition appears in the initial years of life, characteristically with pigmented nevi on exposed parts of the skin. The most dreaded complication is cutaneous cancers, especially Basal cell carcinoma. Ocular involvement is reported in approximately 40% and the median age of onset of ocular symptoms is four years.[1]

Ocular manifestations of XP are primarily ophthalmohelioses those that have been linked to UV exposure of the eyelids, cornea, or lens. These commonly include photophobia, ectropion, symblepharon, corneal opacities, ulcerations and epithelioma of lids among many others. Herein we report four cases with ocular involvement.

CASE REPORT

Case 1

A six years old male child born of consanguineous marriage presented with bilateral ocular swelling and mucopurulent discharge, along with maggots in a necrotic area near lateral canthus of left eye.

Patient also had photoexacerbation over existing skin lesions. Ocular examination of right eye revealed visual acuity finger counting at 1meter. Additionally, ectropion and keratomalacia was noted. Due to difficult eye opening cornea, anterior chamber and disc could not be assessed. Lids were indurated with mucopurulent discharge and bleeding on touch. Ulcerated lesion approximately sized 1×1 cm was noted near lateral canthus extruding maggots (Figure 1).

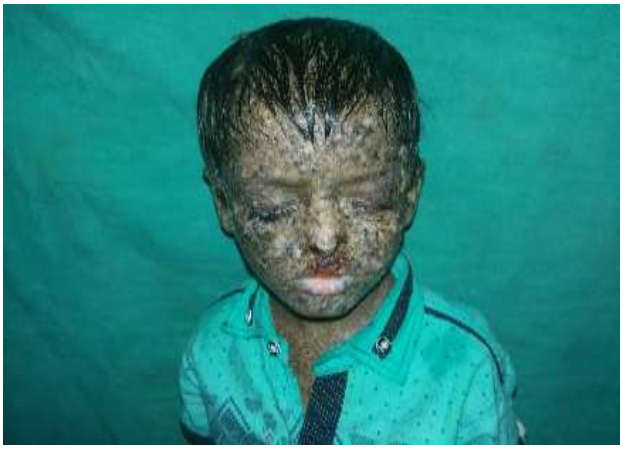

Figure 1: Multiple hyperpigmented macules and papules of variable size over face with single crusted ulcerative lesion over upper lip.

General and systemic examination revealed no significant abnormality. Dermatological examination showed hemorrhagic crusts over both cheeks, left temporal area, nasal alae. Single deep ulcer approximately sized 4×3 cm size with well defined margins and floor covered with granulation tissue was seen over lateral canthus of left eye. Single crusted ulcerative lesion of size 2×2 cm was noted on upper lip involving philtrum area. Multiple hyperpigmented macules and papules of variable size were seen over face, ear, neck, back and bilateral upper and lower limbs. Clinical diagnosis of XP was made.

Routine investigations showed no abnormalities except raised triglyceride levels. CT Orbit and paranasal sinusbc suggested preseptal edema with soft tissue thickening on left side. Histopathological examination from ulcerated lesion over left eyebrow showed keratin material along with parakeratotic sheets of squamous epithelium. Skin biopsy from lip showed stratified squamous epithelium tumour, tumour cells arranged in tubules and cord with degenerated mucin material, surrounding cells had basaloid appearance, so final diagnosis of Basal cell carcinoma was made. Patient was admitted and maggot removal was performed using terpentine oil with daily dressing. He was started on I.V. antibiotics and ulcerative ocular lesion resolved in 15 days. Surgical excision of lip malignant lesion was done under appropriate anesthesia and aseptic precaution. Raised serum lipid profile precluded use of retinoids as chemoprophylaxis.

Case 2

A four year old male child born of consanguinous marriage was admitted with complaints of blackish discoloration over face and neck and asymptomatic lesion over nose. History of photosensitivity was elicited. General and systemic examination revealed no abnormalities. On dermatological examination multiple hyperpigmented macules and dull black papules of variable size were present over forehead both cheeks and nose. Erythema present over medial side of right eye. Single crusted plaque of approximate size 3×2 cm was present over dorsum of nose (Figure 2). On clinical examination diagnosis of XP was reached.

Figure 2: Crusted plaque of approximate size 3×2 cms was present over dorsum of nose.

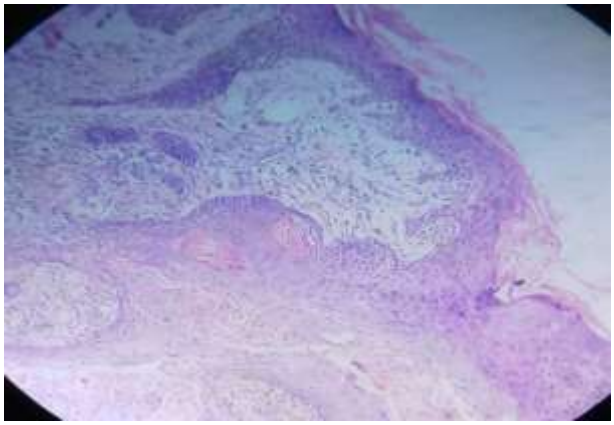

Figure 3: H & E (10 X) stratified squamous epithelium with irregular acanthosis and irregular projection of squamous cells with keratin pearl formation.

Routine investigations were within normal limit. Histopathological examination (Figure 3) performed from crusted plaque over nose showed stratified squamous epithelium with irregular acanthosis and irregular projection of squamous cells with keratin pearl formation suggestive of actinic keratosis.

On interrogation, it was revealed that his younger two siblings also had complaints photophobia. On examination, there were multiple hyperpigmented freckle- like lesions predominantly on sunexposed sites. They are currently under thorough evaluation in view of early changes of opthalmoheliosis.

DISCUSSION

XP is an autosomal recessive disease of defective DNA repair that affects males and females equally and is

frequently symptomatic since childhood. Defects in nucleotide excision repair can lead to three diseases: XP, cockayne syndrome, and trichothiodystrophy. XP and cockayne syndrome both present with photosensitivity and progressive neurological degeneration. XP has a greatly increased risk of sun-induced cancers while cockayne patients have normal cancer risk.[2]

Ocular disease is evident in at least 40% of XP patients, and blepharospasm and photophobia are common symptoms. Eyelid skin changes reflect local skin changes, including erythema, pigmentation, atrophy, and malignant change.[3] Telangiectasias, loss of lashes, and chronic blepharitis are also seen. Atrophic scarred skin may cause ectropion of the lower eyelid and symblepharon. Lower lid loss may result in exposure keratitis, edema, and even corneal ulceration and perforation, corneal opacification. Conjunctival involvement usually includes conjunctivitis, pinguecula, symblepharon, melanosis, and tumors developing from the interpalpebral zone of the limbus. Limbal tumors, especially pterygia, 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, melanoma. Orbital tumors include basal cell carcinomas, squamous cell carcinomas, and melanomas. Eyelid and conjunctival cancers are the most commonly reported. As the posterior segment is protected from UV damage by the cornea and lens, fundus abnormalities are not common; however, choroidal melanoma rarely develops.[4]

In classical XP, the median age of onset of the cutaneous symptoms is between 1 and 2 years. The sun-exposed skin becomes dry, pigmented and parchment-like, with freckle-like hyperpigmented macules. Premalignant actinic keratoses develop at an early age. Patients with XP under 20 years of age have greater than 1000-fold increased risk of cutaneous basal cell or squamous cell carcinoma or malignant melanoma.[5]

As seen in our cases, ocular examination plays crucial role in patients of XP. Continous scrutiny and high index of suspicion is needed to prevent and treat malagnancies as early possible. Although treatment of XP is not satisfactory, children need total protection from sunlight. Even the light coming through windows or from fluorescent bulbs could be harmful.

Use of high protection sunscreens to help prevent malignant transformation of precancerous growths is to be recommended.

Clinical management of XP includes avoidance of sunlight, minimizing UV and cigarette smoke exposure, early excision of skin lesions, and genetic counseling. Oral 13-cis retinoic acid has been shown to reduce the incidence of new cancers in XP patients. Ophthalmic management includes UV-absorbing sunglasses with side shields, artificial tears, intermittent topical steroids, surveillance for ocular neoplasms, and management of complications.

CONCLUSION

XP is a rare disease caused by a defect in DNA repair, often with cutaneous and ocular sun sensitivity, skin pigmentation, multiple skin and eye cancers, and, in some patients, progressive neurodegeneration. Management of XP includes strict photoprotection and close monitoring of premalignant lesions. This case series underlines the importance of ocular examination at every visit to avoid catastrophic changes. It also illustrates the role of the dermatologist in prevention of ocular complications which may severely impact quality of life.

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