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Case Study

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A TALE OF FOREIGN BODY SENSATION TO CARCINOMA-IN-SITU: A CASE REPORT ON XERODERMA PIGMENTOSUM

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ABSTRACT

Xeroderma pigmentosum (XP) is a genetic disorder of childhood presenting with various oculo- cutaneous changes and even malignancies. In this report, we bring forth the case of a 16 year old female, who just thought had foreign body sensation in eye with pigmentation all over the face. On evalauation she was diagnosed as a case of XP with carcinoma-in-situ in conjunctiva and limbus.

KEYWORDS: Xeroderma Pigmentosum, carcinoma-in-situ, xerosis, tylosis, hyperpigmentation.

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare genetic disorder due to mutations in genes responsible for DNA repair, specifically nucleotide excision repair (NER), characterised by severe hypersensitivity to ultraviolet (UV) light. Ocular manifestations include photophobia, conjunctivitis, secondary exposure keratitis with subsequently corneal opacification and vascularization, ectropion, pterygium and neoplasia. The spectrum of neoplasia affecting the ocular surface and adnexa of patients includes squamous cell carcinoma (SCC), basal cell carcinoma (BCC), and malignant melanoma (MM).[1] A preneoplastic spectrum known as conjunctival intraepithelial neoplasia (CIN), classified as mild, moderate or severe can also present secondary to sun exposure with this underlying genetic disorder. In this report, we present a 16 year old female with ocular manifestations of XP.

CASE REPORT

A 16 year old female presented to the OPD with complains of foreign body sensation in both eyes since 7 days; associated with mild redness. No history of associated pain, itching, trauma or drug use or any treatment before was present. No parental consanguity. No similar illness in family. On examination, patient had skin lesions all over the body since birth. There were visible brown, disfiguring spots and patches of different sizes and irregular shapes on face (Figure1). Diagnosis of XP was made by dermatology department. Physical examination showed that patient's general condition and vital signs were normal, with no enlargement of superfi-cial lymph nodes.

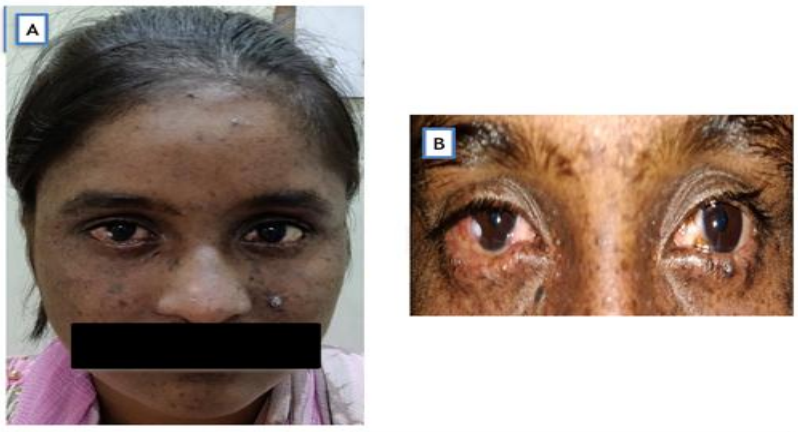

Fig. 1 (a) Gross photograph of patient showing freckles all over the face (b) Bilateral eyes showing various findings.

On ocular examination BCVA was 6/6 OU. Patient was photosensitive. Right eye upper lid was hyperpigmented and excoriated with loss of eyelash in medial 2/3rd of lower lid. There was tylosis with moderate ectropion of lower lid. Limbal and bulbar conjunctival growth, encroaching upon the cornea on temporal side was present. Congestion of conjunctiva was present between 3o'clock to 9o'clock. 2mm of corneal opacity was present between 5o'clock to 7o'clock. Left eye upper lid was hyperpigmented and excoriated. Mild ectropion was present in lower lid. Dark pigmentation was present on supero-nasal and infero-nasal conjunctiva. Cornea was opacified inferiorly (Figure2). Both puncta were visible and patent and Schirmer's test was also normal in both eyes. Neurological examination was within normal limit. Various investigations eg. hemogram, lipid profile, liver and renal function tests, ultrasound abdomen were found to be normal. In right eye wide excision of the conjunctival mass was done.

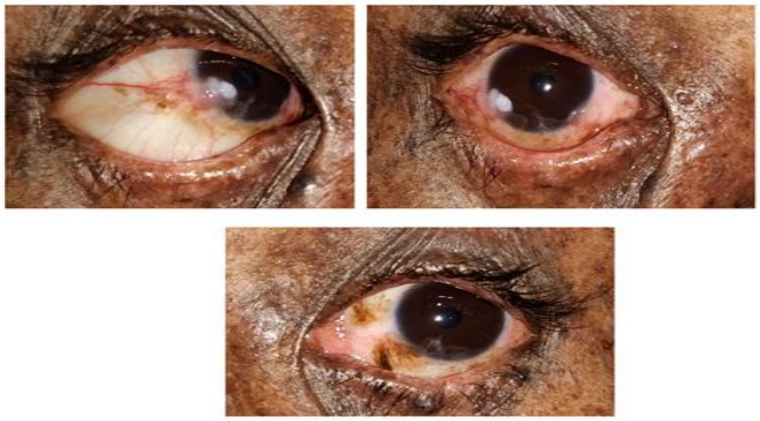

Fig.2(a, b): RE-hyperpigmentation, excoriation, tylosis, loss of eyelashes in medial ⅗ rd of lower lid, conjunctival congestion with a mass encroaching towards cornea and corneal opacity in lower part. (c)LE — hyperpigmentation, excoriation, tylosis, hyperpigmentation in conjunctiva and corneal opacity in lower part.

Histopathology reports (Figure 3) showed nests of squamous epithelial cells arising from the epidermis and extendinginto the dermis. Few large cells with abundant eosinophilic cytoplasm and a large nucleus were seen. All the feature were suggestive of carcinoma-in-situ (grade1) in the lesion.

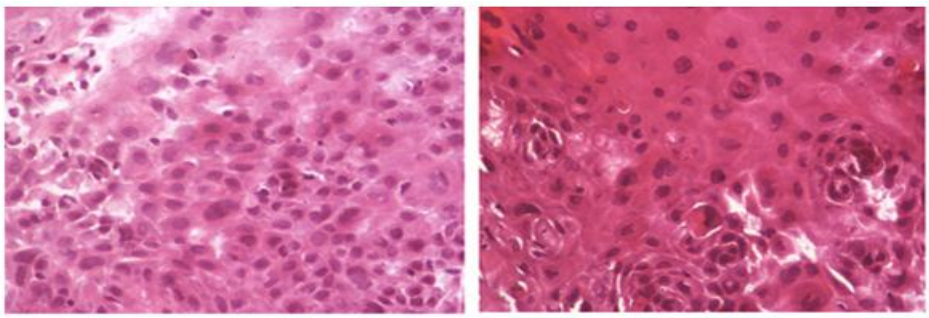

Fig. 3: a, b: Conjunctival lesion (H&E X200) and Limbal lesion (H&E X200) showing nests of squamous epithelial cells arising from the epidermis and few large cells with abundant eosinophilic cytoplasm and a large nucleus.

DISCUSSION

In 1968, Cleaver[2] was first to report that skin cells from patients with XP have an impaired ability to repair ultraviolet radiation induced DNA damage. XP has a prevalence rate of 1:250000.[3] XP being an autosomal recessive disease, the heterozygous parents are clinically normal and both sexes are equally affected. Parents in our case were also clinically normal. Parental consanguinity which is important in any autosomal recessive disorder was not present in our case. The symptoms and signs are usually evident by the end of the first year or the beginning of the second year of age. Our patient had developed the complaints by 10 years of age.

Cutaneous changes in XP include[4] increased photosensitivity, pigmentation, malignant and atrophic

skin lesions. Skin neoplasms range from squamous cell carcinoma, basal cell carcinoma to malignant melanoma.

The earliest ocular symptom of XP is photophobia which is found in 21% of patients, and was seen in this case too as one of the chief complains for presentation. It has been observed that photophobia is more commonly seen in younger individuals[5] than in adults. The reason and mechanism for this is unknown.

The involvement of the lids is seen in more than 80% of reported case and in our case lid freckles and atrophic skin changes of eyelids were seen. Cutaneous freckles and hyperpigmented spots represent clones of individual melanocytes altered by mutations induced by ultraviolet radiation. Due to relative protection of upper lid from ultraviolet radiation from sunlight by the overhanging superior orbital margin, upper lid changes are either absent or much less compared with lower lid.

Conjunctival involvement which includes telangiectasia, xerosis, chronic conjunctival congestion, and pigmentation are most prominently seen in sun exposed interpalpebral fissure and have been reported in 18% of patients. Though no evidence of dry eye was seen in the patient, conjunctival congestion and conjunctival mass were present along with pigmentation of conjunctiva in left eye.

The cornea in XP may show dryness, exposure keratitis, hazyness, band-like nodular keratopathy, scarring, ulceration, and even perforation resulting in corneal opacities and vascularisation. According to Applegate and Ley,[6] DNA damage is involved in the induction of opacification and neovascularisation of the cornea by ultraviolet rays.

Ocular surface neoplasms, including squamous cell carcinoma (most frequent), basal cell carcinoma, and melanoma, occur with a predilection for the limbal area. Our patient had a mass in limbal area extending to cornea which was diagnosed to be CIN grade 1.

Visual impairment in XP patients is attributable to several causes such as corneal opacities, pterygium, tumour invasion from the limbus, and corneal vascularisation. The incidence of visual impairment has been reported to be 12%.[7] But in our case BCVA was not hampered.

General management of XP patients includes protection from ultraviolet radiation, early excision of neoplasms, and genetic counselling. Eye care consists of sunglasses, artificial tears, steroid drops, and bland ointment at night. Surgical treatment includes excision of neoplasms, release of symblepharon, and keratoplasty for corneal opacification. Photophobia and ocular irritation in patients exposed to ultraviolet irradiation are relieved with eye drops containing quinoline derivatives. Oral retinoic acid has also been claimed to be helpful in these patients.

Since, XP patients have potential blinding complications, an ophthalmologist must be involved from the beginning in the care of these patients.

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