

Conjunctival malignant melanoma in xeroderma pigmentosum - a case report

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Abstract Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterised by photosensitivity, pigmentary changes, premature skin ageing and development of various cutaneous and internal malignancies at an early age due to defective nucleotide excision repair following exposure to ultraviolet radiations. Conjunctival malignant melanoma has a very rare association with XP. In this regard, we report a case of a 14-year-old boy suffering from XP with malignant melanoma of conjunctiva.

Key words

Xeroderma pigmentosum, malignant melanoma, conjunctiva.

Introduction

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterised by photosensitivity, pigmentary changes, premature skin ageing, neoplasia and abnormal DNA repair.¹ XP was described in 1874 by Hebra *et al.*² and the term 'xeroderma pigmentosum', meaning pigmented dry skin, was introduced in 1882.³ The basic defect is an abnormal nucleotide excision repair pathway, responsible for the removal and replacement of damaged DNA⁴ which causes defective DNA repair in cells damaged by UV radiation, resulting in photosensitivity and carcinogenesis. In 20% of

the cases, the patients have normal excision repair, but a defective postreplicative repair.⁵ Parents of affected individuals are clinically normal and consanguinity may be seen in about 30% cases.⁶ A variety of tumours like basal cell carcinoma, squamous cell carcinoma, angiosarcoma and keratoacanthoma have been reported with XP with rare association with melanoma and fibrosarcoma. Ocular involvement is seen in 80% of the cases.⁶ We report a case of malignant melanoma of conjunctiva, confirmed histopathologically, in a patient of XP.

Case report

A 14-year-old boy presented with complaints of multiple freckles along with dry and scaly skin all over the body. The lesions were present since 6 months of age. The lesions first started appearing on the nose and later spread all over the body. The patient also had photosensitivity and photophobia. The patient presented with a

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Figure 1 Patient of xeroderma pigmentosum with conjunctival malignant melanoma.



Figure 2 Sister of patient with xeroderma pigmentosum.

swelling over the left lower eyelid since last 2 years and its size was progressively increasing (**Figure 1**) leading to difficulty in opening and closing of the left eye. Similar complaint of XP was also present in patient's sister (**Figure 2**) but there was no history of melanoma in any other member of the family. There was no history of any trauma or foreign body.



Figure 3 Case after excision.

On clinical examination, the patient had multiple hyperpigmented macules (freckles) interspersed with small round atrophic hypopigmented macules and crusted lesions at places. There was conjunctival congestion in the left eye. A hyperpigmented swelling around 16×12×7 mm in dimension was seen over the bulbar conjunctiva. Fundus examination was normal. All routine haematological investigations were within normal range and the CT scan of the orbit and the brain revealed no abnormality. There was no regional lymphadenopathy and no neurological manifestation was reported. Wide excision of the swelling was done and lid reconstruction was done by McGregor transposed flap under bulbar anaesthesia by a plastic surgeon (**Figure 3**). The excised specimen was then sent for histopathological examination which revealed relatively well defined skin lining beneath which vaguely capsulated lesion, consisting of diffuse solid sheets of highly malignant looking cells showing marked pleomorphism with hyperchromatic to vesicular nuclei with prominent nucleoli and large amount of melanin pigment, was seen. The features were suggestive of malignant melanoma. Since the histopathological examination was diagnostic of melanoma, immunochemical markers were not done.

Discussion

XP is a very rare disease with an incidence of approx. 1: 250000 in Europe and USA.⁷ The parents being heterozygotes are normal and healthy while the children are affected. The patients are highly photosensitive, with the skin and eyes highly prone to be damaged by exposure to ultraviolet radiations. The patients of XP may develop neoplasms at a mean age of 8 years as compared to 60 years in normal individuals. Prenatal diagnosis can be made by an analysis of DNA repair of cultured amniotic cells. There is genetic heterogeneity identified by somatic cell fusion studies or complementation groups. There is depletion of Langerhans cells in the skin of XP patients by UV radiations. There are decreased T helper cells to suppressor cell ratio, impaired production of interferons in lymphocytes and reduced natural killer cell activity. In addition to having a greater than 1000-fold risk of multiple skin cancers (basal cell carcinoma, squamous cell carcinoma, and less frequently, melanoma), patients of XP have a 10 to 20-fold increased risk of developing several types of internal cancers.⁸ Several cutaneous melanocytic lesions with the ability to undergo malignant transformation have been identified as possible precursor lesions to others.

Malignant melanoma is a malignant neoplasm of epidermal melanocytes. It is the third most common skin cancer and represents 3-5% of cutaneous malignancies. Ocular neoplasms occurring in XP in order of frequency are squamous cell carcinoma, basal cell carcinoma and melanoma.⁶ The first description of an ocular malignant melanoma was by Greer in 1966.⁹ Among the ocular tissues, the lids, conjunctiva and cornea are predominantly affected because these are exposed to ultraviolet radiations from sunlight.⁶ The development of

conjunctival malignant melanoma in XP patients may support the putative role of sunlight exposure in malignant transformation of conjunctival melanocytes. The case has been reported due to its rare presentation. Malignant melanoma in a patient of XP requires a very early and a proper diagnosis. Such cases are treated by complete excision of tumour and genetic counselling along with medical management.

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