XERODERMA PIGMENTOSUM WITH MALIGNANT MELANOMA AND SQUAMOUS CELL CARCINOMA

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A 25-year-old female with xeroderma pigmentosum since 3 years of age, developed a nodular growth on the left ala of the nose since 4 months. Histopathology revealed malignant melanoma of the nodular variety. A squamous cell carcinoma was also detected at the limbus in the right eye. There were no metastases.

Key words : Xeroderma pigmentosum, Malignant melanoma, Squamous cell carcinoma.

Xeroderma pigmentosum is a rare recessively inherited disorder of defective cellular repair of DNA damaged by ultraviolet rays. It is characterised by photosensitivity, xerosis, pigmentary changes, atrophy and keratoses. Progressive ocular and neurologic degeneration may occur in some cases. There is an increased tendency to develop malignancy in the sun-exposed areas at an early age. Several types of neoplasms are known to occur. We saw association of malignant melanoma in the skin and squamous cell carcinoma in the eye in a case of xeroderma pigmentosum.

Case Report

A 25-year-old female born of non-consanguineous parents had xerosis, freckle-like spots and small, white atrophic macules since the age of 3 years. Lesions were present mainly on the sun-exposed areas. Easy sunburn, photophobia and increased lacrimation were also present for the same duration. The symptoms had become worse in the past 2 years. About 4 months ago, she developed a nodule over the left ala of the nose which bled occasionally. It was firm, pigmented, non-tender, freely mobile, sessile, measured 2×2 cm in diameter, with a lobular surface showing haemorrhagic crusting. There was intense blepharospasm and conjunctival hyperemia in both the eyes. Right eye had a greyish-white nodular growth of 4×4 mm size situated at the limbus on the temporal side encroaching upon the cornea. Vision was limited to counting fingers at 3 metres. Left eye had a vascularised corneal opacity of leucoma grade measuring 5×5 mm overlying the pupil. Only hand movements were perceived in the left eye. Fundoscopy could not be done because of severe blepharospasm.

Mucous membranes, teeth, hair and nails were normal. Lymph nodes were not enlarged. General and systemic examination revealed no abnormality. Sexual and intellectual development were within normal limits. The patient was married since 5 years but had no children. Other members of the family were normal.

Routine investigations were normal. There was no aminoaciduria. Serum proteins, lipids and cholesterol levels were normal. Porphyrin concentrations in blood, urine and faeces were not increased. X-ray of the chest and skull and ultrasound examination of the abdomen did not show evidence of secondary deposits. Histopathological examination of the skin of the forearm showed hyperkeratosis, thinning of the stratum Malpighi, atrophy of the rete pegs and increased melanin pigmentation in the basal cell layer. There was oedema and chronic inflammatory cell infiltration in the upper dermis. This picture was consistent with the diagnosis of xeroderma pigmentosum. Excision biopsy

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of the nodule on the face revealed sheets of spindle-shaped pleomorphic cells with hyperchromatic nucleus and eosinophilic cytoplasm with abundant melanin pigment. The tumour cells were seen infiltrating into the reticular dermis. The adjoining epidermis was normal. This picture was consistent with malignant melanoma of nodular variety and level IV as per Clark and Mc Govern classification.[1] Excision biopsy of the nodule in the eye revealed moderately differentiated squamous cell carcinoma.

Both the neoplasms were excised and skin grafting done on the face.

Comments

Xeroderma pigmentosum was first described by Kaposi in 1870 under the name Kaposi's dermatitis and later in detail by Hebra and Kaposi in 1874.[2] The incidence is about 1 : 250,000.[3] Kraemer et al[4] reviewed published descriptions of 830 patients of xeroderma pigmentosum obtained from a survey of the medical literature from 1874 to 1982 which included 29 cases from India. The median age of the patient was 12 years with a nearly equal sex distribution. Consanguinity of the parents was noted in 31% of the cases.

Among the cutaneous neoplasms in xeroderma pigmentosum, basal cell carcinoma and squamous cell carcinoma are the commonest, while angiosarcoma, fibrosarcoma and keratoacanthoma are rare.[5] The incidence of malignant melanoma has been quoted to be 3%[6] and 5%.[4] In the review done by Kraemer et al,[4] 65% of the malignant melanomas were situated on the face, head and neck. Specific histopathologic types were rarely reported. However, the incidence of the nodular type is quoted to be only 10% of all the malignant melanomas.[1]

Metastases most commonly occur in the regional lymph nodes, but any organ may be affected by the haematogenous spread.[1] Common sites are liver, lung and the central nervous system. The high mortality rate in xeroderma pigmentosum is largely due to metastases, two-thirds of the patients dying before the age of 20 years.[5] Our patient was free from metastases at the time of examination.

Ocular changes include photophobia, blepharospasm, ectropion, entropion, conjunctivitis, keratitis and neoplasms such as epithelioma and melanoma.[7] The incidence of ocular changes varies widely from 40%[4] to 80%.[5] In the review by Kraemer et al, 11% of the patients had ocular neoplasms (excluding those of eyelids). These occurred most frequently at the limbus and were predominantly squamous cell carcinomas. Among 64 patients in whom the age of onset of the first ocular neoplasm was reported, 50% of the neoplasms occurred by 11 years of age. Our patient showed similar features although the exact age of onset of the neoplasm could not be ascertained.

Neurological abnormalities may occur in a few cases. These include progressive mental deterioration, areflexia, impaired speech and hearing, microcephaly and convulsive disorders. Dwarfism and immature sexual development may also be present. Our patient did not have such features. The other abnormalities described in association with this disorder are hyperlipoproteinemia, hypercholesterolemia, aminoaciduria, glucose-6-phosphate dehydrogenase deficiency, higher incidence of internal neoplasms[4] and immune defects.[8,9]

There is no specific treatment for this disorder. Constant protection from sunlight and use of sunscreeners may help to prevent the development of neoplasms.

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