XERODERMA PIGMENTOSUM
(Case reports)

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Summary

Xeroderma Pigmentosum (XDP) is a hereditary disorder characterised by early development of pigmentation, atrophy, keratoses and carcinomas occurring predominantly on light exposed skin. The disease is often fatal before the age of 20 years. Survival beyond middle age is sometimes possible in mild cases with adequate treatment[1]. Two patients with XDP aged 60 years and 75 years were admitted in the Skin department of Rajendra Hospital, Patiala. Survival upto 70 years has been reported by Herxheimer in 1947[9]. Survival in XDP up to the age of 75 years is particularly significant in our country with abundant sunshine which is practically impossible to avoid completely.

Xeroderma Pigmentosum is a chronically progressive multisystem disease, first described by Kaposi in 1870 under the name "Kaposi's dermatitis" and later in detail by Hebra and Kaposi in 1874[2]. XDP is seen in both white and coloured races. Its incidence is estimated to be between 1 in 65,000 and 1 in 100,000 of the population. Important clinical features of XDP are freckles of variable size and colour, increased dryness and erythema of skin particularly on the light exposed areas and mucus membranes of the lower lip and conjunctivae, Later covered areas are also involved. Small irregular white atrophic spots, telagiectasiae, angiomata, scars from superficial ulcers and warty keratoses may also be present. Keratoacanthomas, basal cell epitheliomas, squamous cell epitheliomas and melanomas frequently complicate XDP.

Eye changes are seen in about 80% of cases. These include photophobia, ectropion, destruction of lower eyelids, symblepharon and ulceration, corneal opacities and epitheliomata of lids, conjunctivae or cornea.

Small stature, poor physical development, microcephaly and hypogonadism if associated with XDP is known as 'De Sanctis Cacchione' Syndrome first described in 1932[3]. Later it has been reported by a number of authors[4,5].

Lynch et al[6] have stressed the association of XDP with congenital Ichthyosis and malignant melanomas.

In XDP inheritance is determined by an autosomal recessive gene. Cleaver[7], has reported defective repair replication of DNA in fibroblasts of patients with XDP. Alterations in the DNA might

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predispose to malignant changes in the surviving cells.

Relationship between DNA repair and carcinogenesis is not a simple cause and effect phenomenon, since malignant cells (Hela cells) have ability to repair DNA damage. Further studies in XDP and other hereditary disorders can elucidate the role of DNA and repair mechanism in carcinogenesis. There is probably some linkage between XDP and ABO blood group.

Metabolic abnormalities like amino-aciduria, elevation of serum copper, decrease of blood glutathione, reduced xosteroid excretion and hypergamma-globulinaemias have been reported by various workers.

Case Reports

Case No. 1: A 60 years old female patient was admitted to the Skin ward with history of an ulcer on the nose of 1 year's duration. She complained of pigmentation, dryness of skin and photo sensitivity since the age of eight years. Patient had ulcers similar to the present one 12 years and 20 years earlier. These had been successfully treated. One of the patient's brothers had a similar skin problem.

General and systemic examination revealed no abnormality. Skin showed hyperpigmented spots varying from pin point to $\frac{1}{2}$ cm. distributed all over the body and predominantly on the face. Similar lesions were present on mucus membranes, scalp, palms and soles. Small atrophic spots and keratotic lesions were intermingled with the hyperpigmented spots. A depigmented patch, result of an old burn was present on foot and a keloid on the right arm. An ulcer 3 cm. × 2 cm. was present on the right side of the nose near the inner canthus of right eye. Margin of the ulcer was well defined, base fixed and floor covered with pale granulation tissue. Hair, nails and teeth were normal.

Clinical diagnosis of xeroderma pigmentosum and basal cell epithelioma were made.

Investigations

Routine investigations showed no abnormalities, Blood group – A

Skin biopsy showed hyperkeratosis, thinning of the stratum malpighi and atrophy of rete pegs. There was oedema of superficial dermis. Melanin pigmentation of basal cell layer, melanophores containing pigment in superficial dermis and chronic inflammatory cells in superficial dermis were also seen. This picture was consistent with a histological diagnosis of XDP.

Biopsy of ulcer showed features of basal cell epithelioma.

Case No. 2

A 75 years old male patient was admitted in skin ward with the complaints of photosensitivity and photophobia since early childhood. From the age of 4 years patient had pigmented spots of variable shape and size on the entire skin including the mucous membranes but predominantly on the face and dorsa of hands. He also had white atrophic spots intermingled with hyperpigmented spots. An year prior to his admission patient had noticed two painless nodules, one on the back and the second in the right axilla. He had been operated for similar nodules many times in the past.

General and systemic examination revealed no abnormalities.

Local Examination

Skin showed brown to black pigmented spots of variable size all over the body, predominantly on the face and dorsa of hands. Atrophic white spots were also present. There was one nodule 1 cm. × 1 cm. in size on the back which was firm and fixed to the skin and underlying structures (Fig. Page

No. 345). A second in the right axilla was 2" in diameter and not tender.

The eyes showed corneal and lenticular opacities with keratoconjunctivitis. Fundus could not be examined due to opacities. Hair and nails were normal.

Skin biopsy showed slight hyperkeratosis, thinning of stratum malpighi and atrophy of rete pegs. Oedema and mild inflammatory reaction were present in superficial dermis. Histology was consistent with late stage of XDP.

Biopsy of nodule on back showed features of basal cell epithelioma.

Biopsy of nodule in axilla report could not be traced.

Discussion

Xeroderma Pigmentosum is fatal in two thirds by the age of 20 years as reported by Gongero[8] and most of the cases die by the age of 40 years. However, Herxheimer[9] reported a case surviving upto 70 years. Most of the patients die either due to malignancy and metastasis or due to abnormal susceptibility to infections. The 2 patients with XDP presented in this paper have also survived for an unusually long time. The longevity in our patients is particularly interesting since they have lived all along in a tropical country with constant sun exposure. Prompt and effective management of malignant tumours as and when they appeared would have contributed a great deal to this long survival.

Complete protection from sun exposure, use of sun screening agents, prompt surgical excision of malignant tumours are all necessary in the management of patients with XDP. Fluorouracil locally is useful in treating malignant tumours.

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