A RARE MANIFESTATION OF XERODERMA PIGMENTOSUM

(A case report)

By

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Xeroderma pigmentosum; the incidence[13] of which has been estimated at between 1: 65,000 and 1: 100,000 of the population, is a rare genodermatosis, a single autosomal recessive character due to a single gene. Two or more members of the family may be affected. History of consanguinity is reported. It most often begins early in life, although it has been observed first in adults and even at an advanced age.

The essential abnormality appears to be increased sensitivity to sunlight between wave lengths from 2800 to 3100 Å

Metabolic defects in the form of aminoaciduria, impaired tyrosine excretion, raised levels of serum glutamic acid transaminases and hyper globulinaemia have been reported but their significance cannot yet be evaluated.

Xeroderma pigmentosum has been reported in many races and contrary to the earlier belief it appears to be no less common in those with heavy pigmentation and is therefore not rare in India,[2,4,12,14,15].

REVIEW OF LITERATURE

The first case of xeroderma pigmentsum was recorded by Kaposi[8] in 1870. J. C. White[7] reported a patient of xeroderma pigmentosum who lived for 45 years. Buermann and Gougerot[3] reported the case of a patient getting this disease at the age of 67 years. Loewenthal[10], King and Hamilton[9] reported occurrence of xeorderma pigmentosum in negroes, a race presumed to be immune to this disease. These authors saw a negro lady aged 40 years with xeroderm pigmentosum. Her children were normal. Till 1934, 200 cases have been recorded in the world literature. From then onwards sporadic cases have been reported from time to time.

On reviewing the literature from India, periodical case reports have been published. From 1941 onwards more than 50 cases of xeroderma pigmentosum have been reported by different authors.[1,5,6,11,16].

CASE REPORT

M., 29 years old Hindu male reported at Skin O. P. D. Safdarjang Hospital on 10-3-1967 with complaints of hyperpigmentation all over the body, deformity of nose and loss of scalp hairs with depigmentation for the last 20 years. According to the patient, the disease was preceded by an attack of small pox, when he was 9 years old. He also complained of tumours over the angle of right eye, left side of the nose and chest since last one year, and a tumour over lower lip since six months. Except the tumour over the lower lip which was gradually increasing in size, the

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rest of the tumours were inactive and stationary since last 8 months. No history of photo-phobia or discomfort on exposure to sunlight could be elicited. Neither was any history of consanguinity or freckles or similar complaints in the family.

Local examination

Skin : Hyperpigmented macules and depigmented, discrete, atrophic lesions distributed throughout the body. Mucous memberane was free. Ciatricial type of alopecia on the right side of the scalp present.

Growths : Soft and non-indurated tumours were present on the face, near the angle of right eye, left side of the nose, forehead and chest.

The tumour over the lower lip was firm, black and not attached to deeper structures.

Right sub-mandibular lymphnodes were enlarged, rubbery in consistency and freely mobile. No other lymphnodes enlarged.

Eyes : Bulbar conjunctiva red. Slight ectropian of lower eyelids present. Vision both eyes: normal.

Investigations :

1. Blood Hb 9.6 gm% T. L. C. 9200/cumm D. L. C. P-58%, L-38%, M-2%, E-2%. B. S. R. 28 mm/1st hour. 2. Blood S. T. S. Negative 3. Urine Routine and Microscopic NAD 4. Stool Routine and Microscopic NAD 5. Urine for porphyrins Negative.

biopsy reports :

(1) Two biopsies from hyperpigmented spots 1294/67; 1295/67 Consistent with xeroderma pigmentosum (Fig: 1).

(2) Biopsy from growth on chest 1389/67 Haemangiomota with hyperpigmentation of the basal cell layer of the epidermis. (Fig; 2).

(3) Biopsy from growth over angle of right eye 1712/67 Pigmented intradermal naevus. (Fig: 3).

(4) Biopsy from growth over lower lip 2298/67 suggestive of melanoma. (Fig: 4).

Comments :

The skin is normal at birth; the first symptoms of hyperpigmented lesions chiefly over the exposed parts are noticed between the 6th month and the 3rd year in over 75% of cases, but may appear in very early infancy, in later childhood or in adult life. In our patient, the age of onset of the disease was 9 years with unusual distribution of the lesions sprinkled throughout the body.

Although the disease advances relentlessly and in orderly fashion through its successive stages, the rate of progression is unpredictable and bears no constant relationship to the age of onset. While most cases beginning in early childhood

have reached the tumour stage before the age of twenty, a few run a benign course, and some cases of later onset may develop multiple tumours within a few years. Our patient has lived to the age of 29 years whereas malignant tumours developing in this disease shorten life to teens only.

As the freckles increase progressively in number, telangiectases and small angiomata appear interspersed among them. The first malignant tumour may develop as early as the third or fourth year. Basal cell epithelioma is the commonest variety and very large numbers may appear over the course of years. Squamous epithelioma is also common. Melanomata are not very common and may be multiple; they may lead to early death from wide spread metastasis or may run a benign course, even in adults, although histologically malignant. Other malignant tumours may rarely occur. In our case he had haemangiomatous tumour over the chest and melanoma over the lower lip.

Altough the disease is a photosensitivity dermatosis, there was neither a history of photo-sensitivity nor photophobia in our case. There was no history of the disease or its forme fruste pattern in the family.

Summary.

A case of xeroderma pigmentosum in a patient aged 29 years having multiple haemangiomatous tumours and a rare manifestation of melanoma of lower lip.

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