Case Report

Xeroderma pigmentosum (XP) - A case report
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We report a case of Xeroderma Pigmentosum who attended our BIRDEM Skin OPD with the complaints of multiple painful, ulcerated hyperpigmented nodules and plaques with exudation, crusts and gradually spreading from face to scalp, body for last 12 years. Initially the lesions were macular and started to appear on her sun exposed areas at the age of 5 yrs. The hyperpigmented lesions gradually spreaded and on exposure to sun light there was peeling of her skin with erythema and burning. Later on she developed multiple soft painful nodular swelling with ulceration on face and scalp which were operated several times but results were delayed and improper healing. Chemotherapy and local radiotherapy were also given. She had no ophthalmological or neurological complaints except photophobia. Several biopsy specimens were taken from different sites which confirmed Basal Cell Carcinoma, Squamous Cell Carcinoma, Seborrhoec Keratoses and Solar Keratoses. Different modalities of treatment including curettage and cautery on big lesions, cryotherapy on small lesions and topical application of 5 Fluouracil were given. Improvement was not satisfactory with re-appearance of new lesions, Finally on radiation therapy, the big lesions improved. Our observation is that it's rare disease in our population. We have reviewed the case with all classical complications of Xeroderma Pigmentosum.

Introduction

Xeroderma Pigmentosum (XP) is a rare autosomal recessive disease characterized by photosensitivity, pigmentary changes, premature skin aging, neoplasia and abnormal DNA repair. Some patients with Xeroderma Pigmentosum have, in addition, neurological complications. XP has 8 different (A-G) and XP variant subtypes on the basis of various chromosomal localization. XP occurs with an estimated frequency of 1 :2, 50000 in USA and Europe. It is more common in Japan (1 :40,000). Male and female ratio is almost equal. There is no data regarding incidence of XP in our population.

Fig. 1: Showing large surgical scar, actinic Keratoses as well as multiple hyperpigmented nodules & plaque

Fig. 2 : Showing large crusted ulcer (s.s.c) on the middle of scalp

Fig. 3 : Showing several large hypetpigmented scbo. Kerototic lesions front & below the ear as well as multiple macular hypetpigmented lesions

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A 45 years old non diabetic, normotensive house wife having one son hailing from Dohar, Nowabgonj, Dhaka attended on October 24, 1999 with the complaints of - Multiple painful, ulcerated hyperpigmented nodules and plaques with exudation and crust
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and gradually spreading from face to Scalp, body -for 12 years. She started erythematous maculopapular lesions on face, upper limbs, chest and back at the age of 5 years. There was no burning, no itching or no relation to sun exposure. She started peeling off her skin with erythema and burning while exposed to sun. But there was no eruption or vesicles or blister. The hyperpigmented lesions gradually spreaded without significant complaints.

12 years back, she developed soft painful nodular swelling with ulceration (6x8 cm) on her forehead, which was operated in Holy Family Hospital and discharged with proper healing. After about 4 months, there appeared new lesions on her face and upper part of body. She was operated 4 times on several occasions within 10 years on several sites, which had delayed improper healing. One year after first operation, local radiotherapy was given with total 15 exposures with satisfactory regression.

Then she consulted dermatologist. Many topical medication including antibiotic, antifungal and steroid as well as repeated cryotherapy were given.

On July 16, 1998 she was admitted in a private clinic with multiple, small ulcerated nodules on scalp and body including a big ulcer on right supraauricular region. She was given chemotherapy consisting of MTX (50 mg), Leucovorin (15 mg), Bleomycin (15 mg) on several occasions.

She came to BIRDEM on October 24 1999 with infected, painful multiple nodules with ulceration, swelling, exudation and crust having offensive smell on scalp, face, chest, back and upper arms. She had no ophthalmological or neurological complaints except photophobia. She had positive family history of same disease. Her brother died in the same disease at the age of 50 yrs.

Our provisional diagnosis was Xeroderma Pigmentosum with Basal cell carcinoma, Squamous cell carcinoma, Melanoma, Sebo. Keratoses and Solar Keratoses. Under systemic antibiotic cover 3 biopsy specimens were taken from different sites for histopathological confirmation which revealed Solar Karatoses, Pigmented Sebo. keratoses, and Pigmented B.C.C. Previously, S.C.C was confirmed histologically from operated specimen.

We advised her to avoid sun exposure with use of sun blocking cream for lifelong. 5 Flurouracil was applied topically on Solar Keratotic lesions till clear once. Curettage and cautery was done on big lesions and cryotherapy was given on small lesions. Improvement was not satisfactory with appearance of new lesions. Finally on radiation, the big lesions improved. She was advised for follow up regularly. But she did not attend properly.

Fig. 4 : Showing multiple pigmented maculopapular lesions of varying size and intensity as well as some nodular lesions on the back.

Fig. 5 : Showing basal cell carcinoma cells of different shapes and sizes, The peripheral cell layer of the tumour masses shows a palisade arrangement of the nuclei

Fig. 6 : Showing hyperkeratosis, papillomatosis as well as cystic inclusions of horny materials of seborrheic Keratoses
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Discussion

XP is a hereditary disorder in which photosensitivity is associated with distinct clinical and cellular phenotypes. XP involves DNA repair and replication deficiencies that predispose homozygous individuals to a 1000 fold increase in non melanoma and melanoma cancers on sun exposed skin surface. Genetically different forms of XP have been identified by cell fusion. The classical form of XP ranges from group ‘A’ to ‘G’. Genetic and molecular analyses have revealed that the repair of ultraviolet induced DNA damage is impaired in classical form of XP patients owing to mutations in genes that form part of a DNA repair pathway known as nucleotide excision repair (NER). In contrast the cells belonging to the variant class of XP are NER proficient and are only slightly more sensitive than normal cells to the killing action of UV light radiation.

XP occurs in all races. There is little information on its clinical picture, frequency and types of malignant lesions in individuals of Bangladeshi population. Our common people even the general physicians are not aware about the disease. That is why our patient was late to consult proper clinician. She attended with multiple complications of XP. About her management, different modalities of treatment including aggressive chemotherapy and radiotherapy were applied. But it was difficult to control recurrence of lesions.

Management of patients with XP is based on early diagnosis, life long protection from uv radiation exposure and early detection and treatment of neoplasm. It is advisable to have a night job and consanguinity of marriage should be discouraged.

About XP complementation groups (A-G) and variant, we could not specify the group in our patient. Because the molecular level investigations are not available in our country. However at this moment, proper observation and awareness about XP among general population and physicians should be emphasised.

Reference