GENETICS AND GENODERMATOSES

XERODERMA PIGMENTOSUM- A CASE SERIES OF 4 PATIENTS

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BACKGROUND: Xeroderma pigmentosum (XP) is a rare disorder (1 in 250,000 live births), inherited as autosomal recessive genodermatosis, characterized by photosensitivity, freckly pigmentation, premature skin ageing, telengectasias, warty papillomatous growth and malignant tumor development in later stage. It results from mutation in nucleotide excision repair gene and post replication repair defect.

OBSERVATION: CASE 1- 13yr/Male child presented with mottled pigmentation with crusting lesions all over body. He had multiple large nodular growth over face with rapid, aggressive progression and having central erosions which bled easily on touch. On examination, short stature, cognitive impairment, hearing loss, ocular hyperpigmentation and photophobia were seen. The biopsy was suggestive of squamous cell carcinoma. The elder sister(15yr) had similar complaints with unaffected elder brother(21yr). CASE 2- 23yr/Male presented with diffuse pigmentation over body and nodular swelling over face. On examination, he had short stature, mottled appearance, ocular hyperpigmentation, corneal opacity and photophobia. The biopsy from nodular lesion was suggestive of squamous cell carcinoma. CASE 3- 8yr/male boy presented with brownish black pigmentation over face, neck, arms and legs. On examination, photophobia was present. CASE 4- 12yr/male child presented with diffuse pigmentation over sun-exposed areas. On examination, corneal opacities, photophobia, mental retardation were seen. Younger brother(7 yrs) had similar complaints.

KEY MESSAGE: These cases were presented because of its rare occurrence. Two out of four patients had biopsy proven squamous cell carcinoma. Two patients presented early with no detectable malignancy. One patient survived beyond 20 years of age, which is rare.