GENETICS AND GENODERMATOSES

XERODERMA PIGMENTOSA –A STUDY OF 15 INDIAN FAMILIES AND 21 CASES AND A 5 YEAR FOLLOWUP

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BACKGROUND: Xeroderma Pigmentosa meaning “pigmented dry skin” is a rare autosomal-recessive disease characterized by photosensitivity, pigmentary changes, premature skin ageing and neoplasia associated with abnormal DNA repair.

OBSERVATION: In this case series 21 cases (14 males and 7 females) from western India with mean age of 10.5 years at presentation, clinically diagnosed to have Xeroderma Pigmentosa were collected and followed for a period of 5 years, two patients were above the age of 18 years. H/O consanguinity was present in 12 cases. History of early age of onset was present in all of them with earliest being 7 month of age and latest being 4.5 year of age. Delayed milestones were present in 5 cases. Photosensitivity and multiple dark and light colour lesions were main presenting complaints.. Photophobia was present in all our patients, Ocular involvement was present in 11 of our patients (55%), while lenticular opacity was present in five patients (22%) and loss of vision as a result of limbal carcinoma in 1 patient. Cutaneous neoplasm developed in 8 cases during follow-up. There were two mortality during the follow-up period.
Complete blood counts were within normal limits. Histopathology of the growth revealed trichelemmal carcinoma in one case and basal cell carcinoma in 4 cases. Actinic keratosis in 2 cases. Squamous cell carcinoma in 1 & with limbal carcinoma in 1 case. Traumatic growth over tongue in 1 case.

KEY MESSAGE: 15 cutaneous carcinoma were excised during the follow up, All patients were advised to wear two layers of clothing and cover the face and hands whenever going outdoors with strict sun avoidance and also advised sunscreens over all uncovered areas. Sunglasses with UVB protection and side shields were also advised. Early diagnosis with UV protection and close malignancy surveillance will prevent skin cancers.