GENETICS AND GENODERMATOSES

XERODERMA PIGMENTOSUM IN NORTHWESTERN YEMEN: A CLINICOEPIDEMIOLOGIC STUDY

Mishri Lal Khatri [(1)]

Medipulse Hospital (formerly At Saudi Hospital At Hajjah, Yemen), Hospital, Jodhpur, India [(1)]

Introduction: Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder related to deoxyribonucleic acid (DNA) repair defect, leading to abnormal sensitivity of the skin to ultraviolet light, clinically characterized by the early development of pigmentary changes, atrophy, keratosis, and carcinomas, predominantly on light exposed skin with various oral and ocular changes. Some patients also develop neurologic abnormalities, primary internal neoplasm and leukemia.

Objective: To study the clinicoepidemiologic aspect of XP in Yemen.

Material and methods: All 40 patients (24 male and 16 female from 32 families) treated and followed between 1997 and 2014 were subjected to detailed analysis with the help of a standardized protocol. The diagnosis was based on clinical features and histopathologic data, when needed. The diagnosis of tumors was confirmed by histopathologic examination in all cases.

Results: The median age of onset of initial manifestations was 9.5 months and that of malignant lesions was 7 years. Parents of the patients were not affected but history of consanguinity was recorded in 28 patients. Initial lesions, such as dryness of the skin and freckles on the face were noticed in all patients. In addition erythema of the face with photosensitivity was observed in 21 patients. Premalignant and malignant skin lesions observed later were actinic keratosis in 15 patients, lentigo maligna in 1, SCC in 10, baso-squamous carcinoma in 1. Eyes were affected with SCC in 7 and malignant melanoma in 2 patients. SCC of lip developed in 2 patients and that of tongue in one patient. Judicious use of acetretin in 12 patients showed good result.

Conclusion: XP in Yemen is characterized by a relatively high incidence, high percentage of consanguinity in parents of the patients, early onset of initial manifestations and that of malignant tumors and severe ocular and oral lesions.