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Clinico-pathological Study of Xeroderma Pigmentosa: A Case Series of Eight Cases

Authors : Kakali Roy, Sahana P. Raju, Subhra Dhar, Sandipan Dhar

Abstract : Introduction: Xeroderma pigmentosa (XP) is a rare inherited (autosomal recessive) disease resulting from impairment in DNA repair that involves recognition and repair of ultraviolet radiation (UVR) induced DNA damage in the nucleotide excision repair pathway. Which results in increased photosensitivity, UVR induced damage to skin and eye, increased susceptibility of skin and ocular cancer, and progressive neurodegeneration in some patients. XP is present worldwide, with higher incidence in areas having frequent consanguinity. Being extremely rare, there is limited literature on XP and associated complications. Here, the clinico-pathological experience (spectrum of clinical presentation, histopathological findings of malignant skin lesions, and progression) of managing 8 cases of XP is presented. Methodology: A retrospective study was conducted in a pediatric tertiary care hospital in eastern India during a ten-year period from 2013 to 2022. A clinical diagnosis was made based on severe sun burn or premature photo-aging and/or onset of cutaneous malignancies at early age (1st decade) in background of consanguinity and autosomal recessive inheritance pattern in family. Results: The mean age of presentation was 1.2 years (range of 7month-3years), while three children presented during their infancy. Male to female ratio was 5:3, and all were born of consanguineous marriage. They presented with dermatological manifestations (100%) followed by ophthalmic (75%) and/or neurological symptoms (25%). Patients had normal skin at birth but soon developed extreme sensitivity to UVR in the form of exaggerated sun tanning, burning, and blistering on minimal sun exposure, followed by abnormal skin pigmentation like freckles and lentiginosis. Subsequently, over time there was progressive xerosis, atrophy, wrinkling, and poikiloderma. Six patients had varied degree of ocular involvement, while three of them had severe manifestation, including madarosis, tylosis, ectropion, Lagopthalmos, Pthysis bulbi, clouding and scarring of the cornea with complete or partial loss of vision, and ophthalmic malignancies. 50% (n=4) cases had skin and ocular pre-malignant (actinic keratosis) and malignant lesions, including melanoma and non melanoma skin cancer (NMSC) like squamous cell carcinoma (SCC) and basal cell carcinoma (BCC) in their early childhood. One patient had simultaneous occurrence of multiple malignancies together (SCC, BCC, and melanoma). Subnormal intelligence was noticed as neurological feature, and none had sensory neural hearing loss, microcephaly, neuroregression, or neurdeficit. All the patients had been being managed by a multidisciplinary team of pediatricians, dermatologists, ophthalmologists, neurologists and psychiatrists. Conclusion: Although till date there is no complete cure for XP and the disease is ultimately fatal. But increased awareness, early diagnosis followed by persistent vigorous protection from UVR, and regular screening for early detection of malignancies along with psychological support can drastically improve patients' quality of life and life expectancy. Further research is required on formulating optimal management of XP, specifically the role and possibilities of gene therapy in XP.

Keywords : childhood malignancies, dermato-pathological findings, eastern India, Xeroderma pigmentosa
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