VOLUME-6 | ISSUE-4 | APRIL - 2017 • ISSN No 2277 - 8179 | IF : 4.176 | IC Value : 78.46

Xeroderma pigmentosa Syndrome with Squamous cell carcinoma A NOT SO COMMON ASSOCIATION IN YOUNGER AGE PATIENTS

General Surgery

KEYWORDS: Xeroderma pigmentosa, squamous cell carcinoma

Dr. Priya Dhandore — M.S. General surgery F.M.A.S., Assistant Professor, Department of General surgery Government Medical College, Pandharpur Road Miraj – 416410 - Maharashtra

Dr. N. N. Hombalkar — M.S. D.N.B. MCh – PEDIATRIC SURGERY, Associate Professor and Unit in-charge of pediatric surgery, Department of General surgery, Government Medical College, Pandharpur Road Miraj – 416410 - Maharashtra

ABSTRACT

Xeroderma pigmentosa (XP) with cutaneous malignancies in the form of basal cell carcinoma are common. But squamous cell carcinoma of skin has been reported rarely. We report 2 cases of squamous cell carcinoma patients with xeroderma pigmentosa syndrome who presented with ulceration over the nose.

SUMMARY

Xeroderma pigmentosum (XP) is a heterogeneous group of genetic diseases which results from faulty DNA repair mechanism. It is a rare autosomal recessive disease characterized by photosensitivity, pigmentary changes, premature skin aging, and cutaneous neoplasia. In patients with XP, the mean age for skin cancer is 8 years compared to 60 years in the healthy individuals. The most common types of cancer found in XP patients are basal cell carcinoma and SCC, mostly involving the face, head, and neck. Herein we report 2 cases of XP with Squamous celled carcinoma occurring at younger age

CASE REPORT

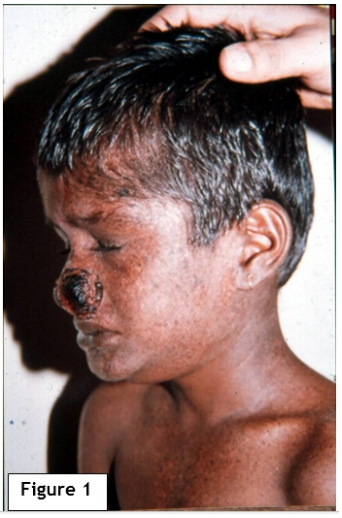

Figure 1

Case 1; A 5-year-old boy belonging to shepherd community presented with an ulcerative lesion on the nose which was not healing. Patient had associated complaints of photophobia, recurrent reddening of eyes and skin pigmentation. He had no history of recurrent skin ulceration. He was born normal with no skin lesions (FIGURE 1). Family history was not contributory. Drug history was insignificant. He achieved developmental milestones according to age and there was no mental retardation. On examination, he had a single non-tender ulceroproliferative lesion on the left nasal ala, circular in shape, around 3 cm in diameter, extending from the left nasolabial sulcus posteriorly and falling short of the tip of the nose anteriorly. The proliferative lesion was hanging below the left nasal alar margin. Lesion showed areas of necrosis and scabbing. General examination revealed generalized skin pigmentation and freckling. There was conjunctival congestion and severe photophobia. Eye examination revealed keratitis. Mental status examination ruled out mental retardation. Systemic examination was within normal limits. Biopsy of the lesion was performed which revealed squamous cell carcinoma. The child was offered definitive treatment in the form of wide local excision and reconstruction. Parents refused to any further treatment.

Case 2; A 5 Years Boy was Presented With ulcero proliferative lesion over the tip of the nose. He had associated complaints of photophobia, recurrent redness of eyes and skin pigmentation. The skin pigmentation and photophobia were worsening over period of time. The ulcero-proliferative lesion was circular and was about 2 to2.5 cm in diameter cm in dimensions with typical rolled out edges (FIGURE 2). It was bleeding on touch. There were many small lesions surrounding the main lesion one such lesion was at the left nasolabial fold and one was in the left preauricular region. Regional lymph nodes were not involved. General examination showed generalized skin pigmentation and severe freckling. There was conjunctival congestion and severe photophobia. Mental status examination was normal. Systemic examination was within normal limits. A diagnosis of XP with cutaneous malignancy was made and the nasal lesion was excised with adequate margins with primary reconstruction. Remainder lesions were also excised. The histopathology of the nasal lesion was squamous celled carcinoma with free margins. The patient was discharged on Day 10 after surgery and was advised about XP protection measures. The child was again seen in Pediatric surgery OPD after 3 months with recurrence of the nasal lesion with 2-3 preauricular lymph nodes. The lymph nodal FNAC suggested inflammatory enlargement. The patient again was operated for wide local excision and forehead flap reconstruction. The child was alright for 8 months after second surgery when last seen in OPD and was lost to follow up after that.

DISCUSSION

Xeroderma pigmentosa was recognised in late 1800 by Maritz Kaposi.[1] Worldwide, the incidence of xeroderma pigmentosa is approximately 1 in 250,000 population.[4] In Japan the incidence is higher approaching 1 case per 40,000 population. Basal cell carcinoma is found to be associated with XP in majority of the reported cases in Indian literature.[2- 3] The basic defect in XP is in nucleotide excision repair (NER), leading to deficient repair of DNA damaged by UV radiation. Two types of NER exist: global genome (GG-NER) and transcription coupled (TC-NER). These genes play key roles in GG-NER and TC-NER. Both forms of NER include a damage-sensing phase, following detection of DNA damage, an open complex is formed. Subsequently, the damaged DNA is removed. The resulting gap is filled in with new DNA by the action of polymerases. There are 10 genetic complementation groups; while one group exhibits defective, post replication repair (XP variant), nine are deficient in excision repair (XP group A-I). Owing to impaired ability to repair, defective or damaged DNA is retained. Retention of the damaged DNA leads to heritable chromosomal mutation and cell death, which possibly cause neoplastic and atrophic clinical abnormalities.[8]

The disease typically passes through 3 stages. The skin is healthy at birth. The first stage makes its appearance after the age of 6 months. This stage is characterized by diffuse erythema, scaling, and freckle

like areas of increased pigmentation. The second stage is characterized by skin atrophy, telangiectasias, and mottled hyperpigmentation and hypopigmentation, giving rise to an appearance similar to that of chronic radiodermatitis. The third stage is heralded by the appearance of numerous malignancies, including squamous cell carcinomas, malignant melanoma, basal cell carcinoma, and fibrosarcoma.

In patients with XP, the mean age for skin cancer is 8 years compared to 60 years in the healthy individuals. Actinic damage occurs between 1 and 2 years. The most common types of cancer found in XP patients are basal cell carcinoma and Squamous celled carcinoma, or in combination mostly involving the face, head, and neck. Isolated Squamous celled carcinoma in less than 8 years have been reported but are not very common

Skin changes are noticed between 12-36 months of age in 75% cases. The cutaneous stages are well defined and similar cutaneous changes are also observed in porphyria and aminoaciduria, however no humoral substances have been demonstrated in xeroderma pigmentosa.

Ocular manifestations include photophobia as the earliest symptom which is a feature of keratitis. Other ocular complications include exposure keratitis, vascularization, ulceration, nodular dystrophy and uveitis.

Neurologic defects are detected in 20-30% of patients with XP. Microcephaly, delayed motor development, dementia, sensorineural deafness are common disorders.[6]

The disease is a progressive one and accelerated degeneration of skin, eyes and nervous system takes place. If left untreated most of the patients die of skin malignancies before the completion of second decade. The course of the disease can be modified by appropriate preventive measures against exposure to sunlight. Prophylactic treatment with topical application (titanium dioxide cream, para-amino benzoic acid in alcohol) may be administered early**.**

Amniocentesis and in vitro cell culture and detection of defective DNA repair may suggest prenatal diagnosis in high risk population.[7]

Cutaneous neoplasia are generally treated with electrodesiccation and curettage or by wide local excision and reconstruction.

IMAGES

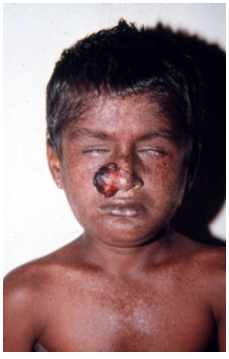

FIGURE 1

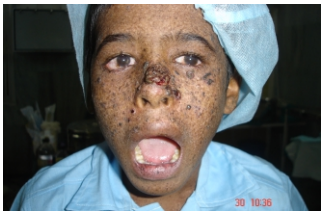

FIGURE 1

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