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BASOSQUAMOUS CARCINOMA IN XERODERMA PIGMENTOSUM - A RARE CASE REPORT

Pathology

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ABSTRACT

Xeroderma pigmentosum (XP), meaning parchment skin and pigmentary disturbance, is a rare and mostly autosomal recessive genetic disorder. The frequency of this disease in the general population of the U.S.A. is 1:250,000. It is characterized by a genetic defect in DNA repair. The consequence is a high incidence of skin cancers on sun-exposed cutaneous surfaces of affected children. First lesions appear in the first years of life: telangiectasia, actinic keratosis and keratoacanthomas. Squamous cell carcinoma , basal cell carcinoma, malignant melanoma are the most frequent neoplasms and rarely fibrosarcoma.We hereby report a rare case of 20 year old male affected by xeroderma pigmentosum, with manifesting as unusual skin malignancy- basosquamous carcinoma.

KEYWORDS

Xeroderma pigmentosum, Basosquamous cell carcinoma, Histopathology.

Introduction:

Xeroderma pigmentosum (XP), meaning parchmentskin and pigmentary disturbance, is a rare and autosomal recessive genetic disorder that was originally named by two dermatologists, the Austrian Ferdinand Ritter von Hebra and his Hungarian son-in-law Moritz Kaposi in 1874 and 1883. [1] XP is characterised by mucocutaneous and ocular hypersensitivity to UV radiation.The basic defect underlying this condition is nucleotide excision repair defect(NER) leading to defective repair of DNA damage by UV radiation [2,3] . The frequency of this disease in the general population of the U.S.A. is 1:250,000. Notably, it is much higher in Japan and other countries. Frequent reports have emanated from other countries including Europe, Egypt, Israel, Korea, China, India and Pakistan [4]. About 50% of persons with xeroderma pigmentosum experience acute sun burn on minimal exposure to UV radiation and tend to develop neurological abnormalities. Cutaneous signs and symptoms usually emerge in children under the age of 20 years. Upto 60% of persons with xeroderma pigmentosum will eventually develop skin cancer.[5]

We hereby report a case of 20 year old male affected by xeroderma pigmentosum, with manifesting as unusual skin malignanacy-basosquamous carcinoma, which has been rarely reported previously.

Case report:

A 20 year old male, 3rd order child, born out of non consanguineous marriage, presented to L.G.Hospital, Maninagar, with the chief complaints of nodular lesion on nose since six months that progressed into ulceroproliferative lesion. Patient has hyperpigmentation over sun exposed areas like face, neck, upper chest, upper extremities and nape of neck since six months after birth. Patient also complained of photosensitivity on exposed to sunlight. He had ocular problems in right eye- photophobia, redness and conjunctivitis since one month. There was no history of any skin surgery in the past. Hairs and nails were normal & no any skeletal abnormality. Family history is that his elder two sisters are also suffering from xeroderma pigmentosum and hyperpigmented skin lesions.

On examination, ulceronodular lesion seen on tip of the nose measuring 1.5x1 cm. Margin of the lesion is hyperpigmented with rolled out irregular border. Ulcer is fixed to the surrounding skin and ulcer base is necrotic.

Biopsy of the lesion of the nose received from dermatology department and sent to histopathology department.

The specimen is processed by Routine paraffin technique. Tissue section was prepared and stained with Hematoxylin & Eosin stain (H&E stain).

HISTOPATHOLOGY

GROSS EXAMINATION: Specimen consists of brownish soft tissue portion measuring 0.5x0.5 cm. On cut section, firm and whitish.

MICROSCOPIC EXAMINATION: Section shows tumor to be composed of nests of basaloid cells with peripheral nuclear palisading. There is also presence of strands of epithelial cells forming nodules with a lace-like pattern and adenoid pattern. The cells have moderate pleomorphic, densly hyperchromatic nuclei with scanty cytoplasm. Focal melanin pigment is seen. Mitosis is also present. Associated conventional squamous cell carcinoma is seen with keratin pearl formation. All surgical margins are involved by tumor tissue.

DIAGNOSIS is Basosquamous carcinoma.

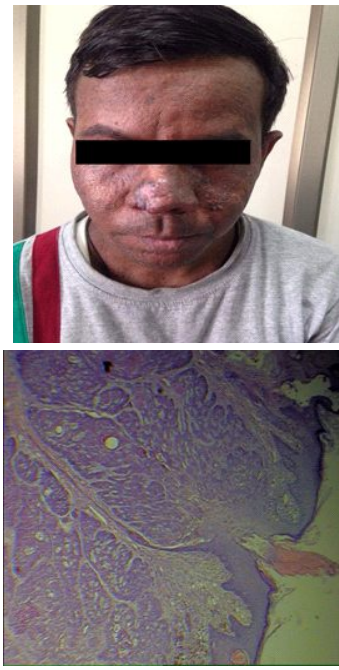

Fig. 1: Basosquamous carcinoma (4X-HE)

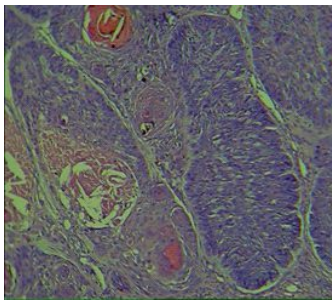

Fig. 2 : Basosquamous carcinoma (10X-HE) –Shows features of both basal cell carcinoma and squamous cell carcinoma. Island of cells with peripheral palisading of basaloid cells. The cells in the central zone showing transition to squamous differentiation. The cells are large with eosinophilic cytoplasm and keratin pearl formation.

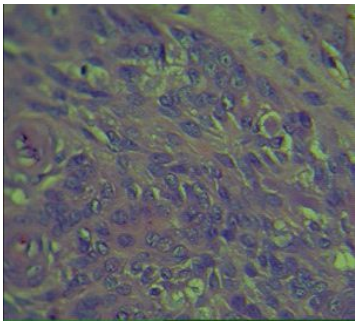

***Fig. 3:* Basosquamous carcinoma (40X-HE) – The cells are large showing hyperchromatic pleomorphic nuclei with mitosis**

Discussion:

Xeroderma pigmentosum, an autosomal recessive disorder , excessive solar damage to the skin develops at an early age. Consequently the lesion occur chiefly in areas of the skin that habitually are exposed to sunlight. [6] The mean age at diagnosis of xeroderma pigmentosum associated skin cancer is 8 years, 50 years younger than in general population. Xeroderma pigmentosum patients below 2 years of age have more than 1000 fold increase risk of developing skin cancer. The most common types of cancer found in xeroderma patients are BCC and SCC , mainly occurring in head, face and neck.Melanomas occur in one fourth of cases.[7]

Clinical hallmarks of xeroderma pigmentosum

- Severe photosensitivity (painful sunburns in early childhood)
- Poikiloderma
- Dryness (xerosis)
- Premature skin aging
- Malignant tumors (squamous cell cancers, basal cell cancers and melanoma),most often on face, head and neck

The nonmelanoma skin cancers (NMSC), which account for mainly basal cell carcinomas (BCC) and squamous cell carcinomas (SCC) develop as multiple primary tumors in XP patients. NMSC are derived from the basal layer of the epidermis with BCC occurring in the hair-growing epithelium whereas SCC derives from inter-follicular cells. BCC arise de novo as slow growing, locally invasive tumors which metastasize rarely. SCC which develop with a multistep progression, are often derived from precancerous skin lesions, actinic keratoses (AK) and tend to be more aggressive and frequently metastasize. [8]

Baso-squamous carcinoma. (BSC) -

BSC is a rare tumor with an incidence of less than 2%. 97% of the tumors were located on the head and neck. Face and ears being the commonest sites.[9] Molecular etiology have been linked to Human papilloma virus in lower animals. [10] Metastases of BSC are known to occur many years after identification of the primary tumor. These tumors are radioresistant. Hence such patients should undergo regular follow-up as long as possible. BSC has biological aggressiveness intermediate between basal cell and squamous cell carcinomas. Authors have described BSC as a wolf in sheep's clothing. A male gender, large size (> 20mm), positive surgicalresection margins and lymphatic or perineural invasion are indicators for the aggressiveness of basosquamous carcinoma.The 5-year survival rate is estimated to be 17.5%.[9] Ocular manifestations include photophobia as the earliest symptoms, which is a feature of keratitis:This was seen in our case. Other ocular complications include exposure keratitis, vascularisation, ulceration, nodular dystrophy and uveitis. Neurologic defect are detected in 20-30% of patients with Xeroderma pigmentosum.Microcephaly, delayed motor development,sensory neural deafness are commonly seen. However in our case ,these were not there.[11] Currently there is no specific treatment for Xeroderma pigmentosum. Managment involves preventing damage and dealing with damage tissue at the earliest.Total protection from UV light greatly improves the prognosis and reduces skin changes and cancers.[12]

Conclusion:

Basosquamous cell carcinoma (BSC) is a rare cutaneous neoplasm with features of both basal (BCC) and squamous cell carcinoma (SCC) associated with xeroderma pigmentosum which has a potential for aggressive infiltration and destruction. This case was presented to create awareness among treating physicians and surgeons about this rare condition and importance of early detection and prevention of UV rays induced skin damage.

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