

## Xeroderma Pigmentosa with Squamous Cell Carcinoma of Lower Lip

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Diffuse pigmentation on lower limbs



Diffuse pigmentation on upper limbs



Squamous cell carcinoma growth on lower lip



Diffuse pigmentation over Abdomen



Diffuse pigmentation on nails



After resection of Growth

Here we report a rare case of 17 years old girl who presented with pigmentation all over the body and mass over the lower lip. There was also history of gradual diminision of vision and photophobia. The pigmentation classically described as freckles varied in colour from light brown and were distributed evenly all over the body involving the face, neck, upper limbs, lower limbs and the trunk. There was associated dryness of the skin.

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Ophthalmic examination revealed bilateral corneal opacities. A cauliflower like growth was seen involving more than 2/3 of the lower lip covered with slough and crust. Neck examination revealed multiple nonspecific lymphadenitis. Rest of the examination including the neurological functions was normal. Biopsy from the mass over the lower lip revealed well differentiated keratinising squamous cell carcinoma. The growth was excised with a wide margin and the defect was reconstructed local flap. Post-opt recovery of the patient was good.

An autosomal recessive rare disease associated with defective DNA repair, it is characterised by severe sun sensitivity resulting in progressive changes of the skin and eyes, often leading to neoplasia early in life<sup>1</sup>. Recognized in late 1800 by Maritz Kaposi, it has been reported worldwide and in all races with an equal sex incidence. It occurs with frequency of approximately one in 250,000 population in Europe and USA<sup>2</sup>.

The disease typically passes through 3 stages. The skin is normal at birth. Typically the first stage appears after age of 6 months, characterised by diffuse erythema, scaling and freckle like areas of increased pigmentation. The second stage is characterised by poikilodermata i.e skin atrophy, telangiectasias, and mottled hyperpigmentation and

hypopigmentation the third stage is heralded by the appearances of numerous malignancies including squamous cell carcinoma, basal cell carcinoma, malignant melanoma and fibrosarcoma. Neoplasia may occur as early as 4-5 years of age and more prevalent in sun exposed area. Ocular problems occur on nearly 80% of cases. Photophobia and conjunctivitis are common early symptom. ectropion and madarosis of lower eyelid and symblepharon and ulceration may occur<sup>3</sup>.

Early diagnosis, lifelong protection from UV exposure and early detection of neoplasms are essential. Strict photoprotection, surgery or chemotherapy of tumours and care of eyes are the mainstay of treatment. Most patients with the full blown syndrome die within the first two decades because of tumor metastases or infections.

**Conflicts of interest :** None reported

**References :**

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