

## **Xeroderma pigmentosum with tongue cancer and neurological manifestations in a 5-year-old child: a case report**

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**ABSTRACT:** Xeroderma pigmentosum is an extremely rare inherited disorder caused by a nucleotide excision repair (NER) defect leading to defective DNA repair after UV radiation-induced DNA damage. The condition is associated with increased risk of cutaneous malignancies, and the manifestations vary across different complementation groups of the disease. Neurologic abnormalities are more frequent in complementation group A, whereas group C has the highest incidence of oral cavity cancer. We describe a case of xeroderma pigmentosum where a 5-year-old child presented with squamous cell carcinoma of tongue and skin, together with neurological and ocular involvement, a constellation of clinical features which has never been reported so early in childhood.

**Keywords:** neurological involvement, squamous cell carcinoma, tongue cancer, xeroderma pigmentosum

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**INTRODUCTION:** Xeroderma pigmentosum is a rare inherited disorder in which the affected individuals are unable to repair the DNA damage caused by UV irradiation. The hallmark of this disorder is extreme photosensitivity of the skin and eyes with premature cutaneous aging and increased frequency of cutaneous malignancies.<sup>[1]</sup> As there is no definite cure, early diagnosis of the condition and regular surveillance is very important for improving

survival rates. Here we report a case of xeroderma pigmentosum with unique constellation of clinical features, namely neurological and ocular involvement along with tongue and skin cancer at a very young age of only five years.

**CASE REPORT:** A 5-year-old boy presented with multiple skin patches and a painless ulcer in his tongue which had gradually increased in size over the previous one year. There was no history of local

trauma preceding the development of the tongue lesion. He was the first child born of a consanguineous marriage. His skin condition was diagnosed as pityriasis versicolor in a rural hospital where he was being treated for the previous two years.

On examination, the child was malnourished and had mild pallor. There was an ulceroproliferative lesion of 2.5 cm x 2 cm size involving the anterior part of the left lateral border of tongue [Figure 1].



**Figure 1: Hyperpigmented macules over the face and ulceroproliferative lesion of the tongue**

The margins of the lesion were everted, and the ulcer bled to touch. There was no leukoplakia or any other lesion in the oral cavity. There were no enlarged cervical lymph nodes. The physical examination also revealed dry skin with numerous hyperpigmented macules involving the face,

neck, upper limbs and chest, and two hyperpigmented papular lesions near the medial canthus of left eye. On enquiry about the skin abnormality, his parents informed that the hyperpigmented skin lesions appeared after the age of two years. Ocular examination revealed photophobia, bilateral

xerophthalmia, corneal vascularization and a pterygium in the left eye [Figure 2]. Neuropsychological evaluation revealed that the child had hyporeflexia and mental

retardation. However, there was no history of seizures and his hearing was normal. The overall clinical features were highly suggestive of xeroderma pigmentosum.



**Figure 2: Pterygium in left eye**

Wide local excision of the lesion on the tongue as well as the two papular lesions below the left eye was performed in the same operation. Histopathology of all the three lesions revealed well-differentiated squamous cell carcinoma. The child is on regular follow up and has not yet developed any new skin or oral neoplasm.

## DISCUSSION

First described by Moriz Kaposi in 1870, xeroderma pigmentosum is an extremely rare autosomal recessive disorder associated with extreme photosensitivity, cutaneous pigmentation, neoplasia, and neurologic degeneration in a subset of patients. <sup>[2],[3]</sup> It is caused by a nucleotide excision repair (NER) defect leading to defective DNA repair after UV radiation-induced DNA damage. <sup>[4]</sup> The incidence is higher in

children with consanguineous birth. [5] Xeroderma pigmentosum is further categorized into different complementation groups according to the DNA reparative capacity. Neurologic abnormalities are most common in complementation group A, whereas group C has the highest incidence of oral squamous cell carcinoma. [6] Neurological manifestations occur in about 30% of patients with xeroderma and can range from sensorineural deafness to areflexia, mental retardation and seizures. [7] The present case has the following unique features:

1) The child has developed oral squamous cell cancer at the age of five years which is the lowest age reported in world literature till date.

2) He has the combination of tongue and skin cancer, plus neurological and ocular involvement at such a young age.

To our knowledge, only three cases of xeroderma pigmentosum with tongue cancer in children have been reported from India, none of whom have such a combination of clinical features as described in our patient. [6],[8] After an extensive literature search, we did not come across a single case report of xeroderma pigmentosum developing both

tongue cancer and neurological manifestations at such an early age. Our case also did not have a positive family history, which probably delayed his diagnosis at the rural hospital and precluded him from taking the necessary precautions of avoiding sun exposure or regular surveillance for cancer.

The present case report also highlights the importance of early diagnosis of this extremely rare inherited condition in children, so that surveillance for skin and oral cancer is possible, thereby helping in early identification of malignant lesions and timely surgical intervention.

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