# *Xeroderma Pigmentosum*: A Rare Disease

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An 8-year-old female, native from a small village in Mozambique, who began symptomatology three years earlier, presented with disseminated hypopigmented and hyperpigmented macules (Fig. 1) as well as nodular lesions on the lower lip, eyelid region, tongue (Fig. 2), and the fifth finger of the left hand (Fig. 3). She reported photophobia and decreased visual acuity during previous years but denied previous sunburn or sun sensitivity. She was an insecure and sad child and she dropped out of school. Her 14-year-old brother had similar skin lesions and a tumor of the lower lip as well as photophobia (Fig. 4). A physical examination revealed generalized freckle-like hypopigmented and hyperpigmented macular lesions, ulcerated tumor with purulent secretion on the fifth finger of the left hand, and multiple tumors in the lower lip, eyelid region, and tongue. Neurological examination was normal. Analytically, she had microcytic and hypochromic anemia. The remaining analytical study was normal, including serology for human immunodeficiency virus. An ophthalmology evaluation reported chronic corneal opacities in both eyes. The radiography of the fifth finger was normal.

The patient underwent antibiotic therapy with crystalline penicillin and the tumor of the left hand was surgically removed (pathology study was not performed). Tumor lesions of the lower lip were surgically removed and confirmed to be epidermoid carcinoma as were the lower lip tumors of her 14-year-old brother.

The presumptive diagnosis was *xeroderma pigmentosum* given the clinical findings and family history. *Xeroderma pigmentosum* is a rare autosomal recessive disorder, which can be found on all continents and in all races and for which there is no cure. It is defined by extreme sensitivity to ultraviolet radiation resulting in sunburn, pigment changes of the skin, and high incidence of skin cancer[1-2]. In addition, patients are susceptible to eye problems, neurodegenerative processes, central nervous system tumors, and other type of tumors. The diagnosis is made clinically and can be confirmed by cellular tests for defective DNA repair[3]. The protection against ultraviolet radiation is mandatory, and the early removal of precancerous lesions can minimize the development of complications. Psychosocial issues need to be addressed[4].

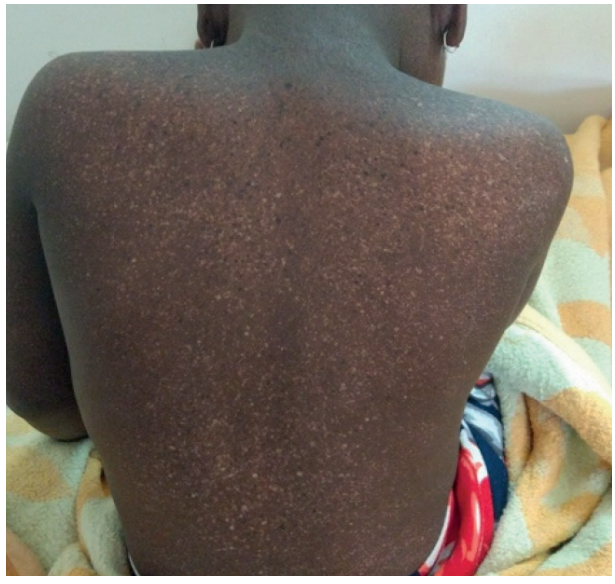

**Figure 1.** Multiple hypo macules on the posterior region of the trunk.

**Figure 2.** Eyelid and tongue tumors.

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**Figure 3.** Ulcerated tumor on the fifth finger of the left hand.

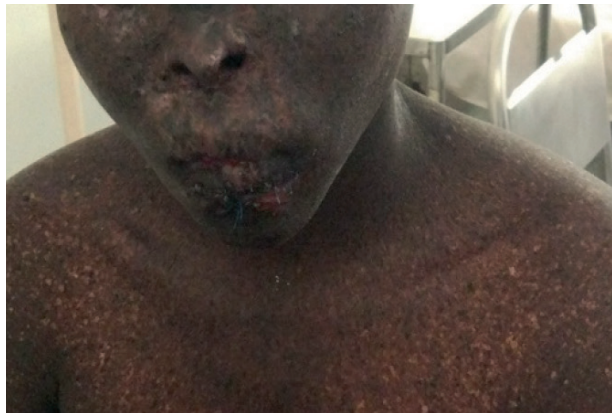

**Figure 4.** Image of the 14-year-old brother with hypo and hyperpigmented lesions on the face and anterior region of the trunk. Small ulcerate lesion on the lower lip.

**Keywords:** Child; Siblings; Xeroderma Pigmentosum/diagnosis

**WHAT THIS REPORT ADDS**

- Xeroderma pigmentosum is a rare entity, and diagnosis is mainly clinical.
- It can be an important cause of morbidity and mortality.
- Psychosocial issues need to be addressed as social isolation is common.
- Early diagnosis and preventive measures are fundamental for the prognosis.

## Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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## Consent for publication

Consent for publication was obtained.

## Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

## References

1. Lehmann AR, Mcgibbon D, Stefanini M. Xeroderma pigmentosum. Orphanet J Rare Dis 2011;6:70. doi: 10.1186/1750-1172-6-70.

2. Black JO. Xeroderma pigmentosum. Head Neck Pathol 2016;10:139-44. doi: 10.1007/s12105-016-0707-8.

3. Haute Autorité de Santé. Xeroderma pigmentosum. National diagnostic and treatment protocol Xeroderma pigmentosum. Saint-Denis La Plaine: HAS; 2007.

4. Genetics Home Reference. Xeroderma pigmentosum [accessed 28 February 2019]. Available at: https://ghr.nlm.nih.gov/condition/xeroderma-pigmentosum