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Xeroderma pigmentosum: when the sun burns the eyes

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Image in medicine

We report the case of a two-month-old male infant from a consanguine marriage. He had pigmented lesions on the skin of the eyelids, the face, and the neck, alternating with areas of depigmentation. An erosive appearance was found on the nose skin and lower labial mucosa. Ophthalmologically, the ocular tracking reflex was present, blepharitis was found with inflammatory palpebral swelling and bilateral ectropion; associated with conjunctival hyperemia, mucous secretions and severe dry eyes with corneal ulceration (exposure keratitis). The rest of the eye exam was normal. The prescribed treatment is based on sun protection, moistening, healing and antibiotic eye drops in both eyes, with close

monitoring. Xeroderma pigmentosum (XP) is a rare genetic disease, with an autosomal recessive transmission, common in the Maghreb due to the high rate of consanguine marriage. The diagnosis of XP is clinical, defined by pigment abnormalities of skin exposed to the sun, skin neoplasia; XP is linked to a defect in the enzymes involved in repairing the oncogenic effects of ultraviolet exposure. Ocular neoplasia is most often located at the limb, cornea and conjunctiva. Prevention of tumors requires protection from sunlight. Surgical removal of tumors and self-healing of skin not exposed to the sun are the most important therapeutic measures. Treatment of complications (hyperemia, infections, and corneal opacification) is symptomatic, waiting for the arrival of the gene therapy.

Figure 1: A) inflammatory palpebral swelling, associated with ectropion of the upper eyelid and conjunctival mucopurulent secretions; B) photograph of the patient showing the presence of pigmented lesions alternating with the areas of depigmentation on the face