Clinical Image

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Xeroderma pigmentosum presenting in two siblings from Uganda

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A 10-year-old female presented to our global health outreach clinic in Uganda in May 2018 for evaluation of progressive pigmentary changes on the face and severe visual impairment. Cutaneous examination revealed a large area of mottled hypopigmentation involving the central nose and upper cutaneous lip (Figure 1). Ocular examination revealed corneal opacities involving the bilateral eyes (Figure 1). The dyspigmentation began during infancy and her vision progressively worsened since age 6. Her 3-year-old brother accompanied her and was noted to have similar dyspigmentation involving the face that had been present since 1 year of age (Figure 2).

Figure 1: A 10-year-old female with mottled hypopigmentation involving the central nose and corneal opacities of the bilateral eyes.

Figure 2: A 3-year-old male with mottled hypopigmentation involving the central forehead, nose, and malar cheeks.

The family stated that both parents had direct relatives with similar cutaneous findings. Based on the clinical findings and family history, both patients were diagnosed with xeroderma pigmentosum (XP).

Xeroderma pigmentosum is an autosomal recessive disorder of DNA repair that is characterized by increased photosensitivity, early pigmentary changes, and UV-induced cutaneous malignancies [1]. Cutaneous manifestations typically present after 6 months of age as persistent erythema, scaling, and ephelides on sun-exposed skin followed by poikiloderma and numerous cutaneous malignancies [1]. There is a 2,000-fold increased risk of melanoma and 10,000-fold increased risk of basal cell carcinoma and squamous cell carcinoma, with a mean onset of malignancy by age 8 [1]. Extracutaneous manifestations may include ocular and neurodevelopmental complications. Common ophthalmologic complications include photophobia, conjunctivitis, ectropion, and symblepharon.

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Neurological deficits may occur including developmental delay, sensorineural hearing loss, hyporeflexia, and ataxia. In severe cases individuals usually die from complications related to metastatic melanoma or invasive squamous cell carcinoma by the age of 20. Diagnosis of XP should be established based upon the clinical findings, family history, and/or confirmatory genetic testing. Sun protection and avoidance with regular cutaneous examinations are essential in the long-term management of XP. Additional chemopreventive measures may be considered including systemic retinoids, topical fluorouracil, and oral nicotinamide [2, 3].

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