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IMAGES IN PAEDIATRICS

Multiple ephelides: Just freckles?

Efélides múltiples: ¿solo pecas?

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We present the case of a boy aged 20 months, of European ancestry, born to consanguineous parents (second cousins). From 1 year post birth, he developed multiple ephelides clustered in areas exposed to sunlight, chiefly in the face (Figs. 1 and 2), leading to the clinical diagnosis of xeroderma pigmentosum (XP). The diagnosis was confirmed with the detection of the homozygous pathogenic variant c.1643_1644delTG (p.Val548Alafs*25) in the *XPC* gene, associated with XP type C. Both parents, who were asymptomatic, carried the variant in a single allele. There was no evidence of ophthalmological or neurologic involvement. The examination of the hair with polarised light microscopy did not reveal abnormalities.

Xeroderma pigmentosum XP is a rare hereditary skin disease (prevalence, 1–2.3/1 000 000) characterised by extreme cellular sensitivity to ultraviolet radiation associated with changes in genes involved in DNA repair.[1] It has an autosomal recessive pattern of inheritance, and there are 8 subtypes: XP type A through G and XP variant.[2,3] It manifests with multiple ephelides from age 1–2 years in areas exposed to sunlight, sunburn after short exposures, premature skin ageing, increased risk of skin cancer (squamous cell/basal cell carcinoma, melanoma) and ophthalmological and neurologic abnormalities.[1–3] The latter are infrequent in XP group C, which is most common in Spain.[2] It can be diagnosed based on clinical features and confirmed with

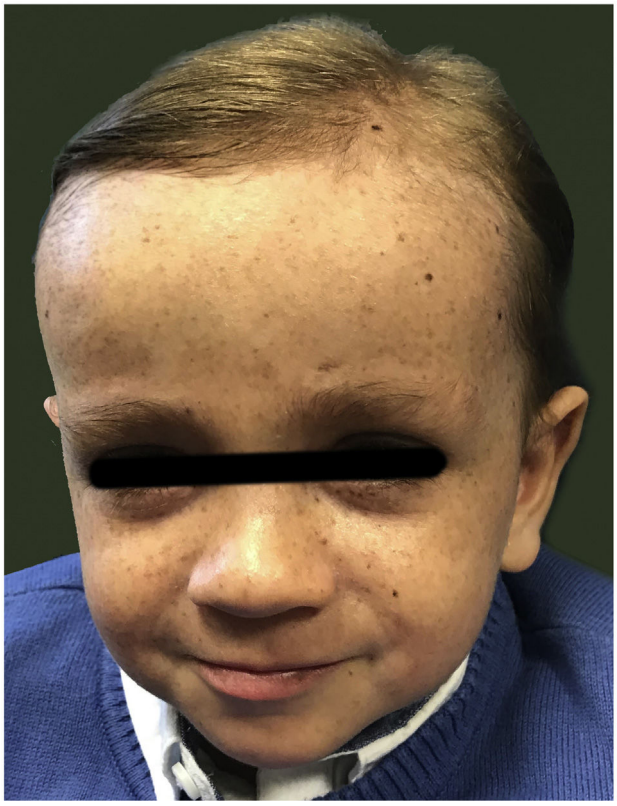

Figure 1 Face of the patient at age 2 years.

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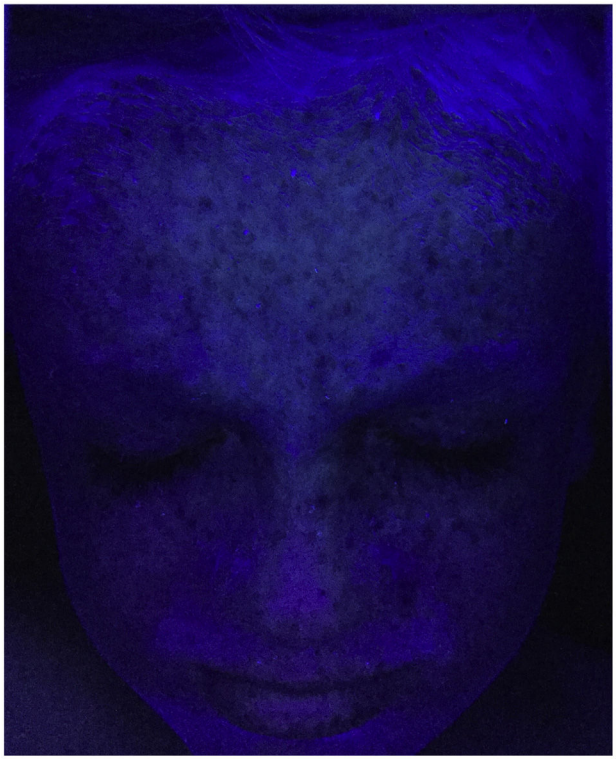

Figure 2 Lesions caused by expossure to sunlight visualised with UV light (courtesy of Javier del Boz González).

molecular methods.[3] The goal of management, based on lifelong avoidance of exposure to ultraviolet radiation, is the prevention and early detection of skin cancer from a young age.[1,3]

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