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PD09 Delving deeper into sunburn
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A 25-year-old woman was referred to a tertiary photodiagnostic service following ‘severe sunburn’ after minutes of sun exposure on an overcast day. She described a prickling sensation on exposed forearms and later developed erythema, vesicles and large unilocular blisters, requiring attendance at accident and emergency and a burns unit. The episode resolved over weeks with exfoliation, but no scarring. With hindsight, she reported five other severe episodes since the age of 7 years, requiring accident and emergency visits and bandaging, so had avidly avoided the sun. She was medically well, on no medications, with no personal or family history of skin cancer. Skin examination was normal, with no skin dyspigmentation or lentigines. Monochromator phototesting showed mild delayed ultraviolet (UV)B and UVA2 sensitivity, with enhanced responses at 48 h. UVB TL-01 minimal erythemal dose, broadband UVA provocation testing, sunscreen photopatch testing, connective tissue disease screen, human leucocyte antigen typing, plasma and red cell porphyrins were normal or negative, and vitamin D levels were markedly deficient. Biopsy of an abnormal phototesting site revealed apoptotic keratinocytes, lymphocytic infiltrate and dermal oedema. Due to the severity of the ‘sunburn’ episodes, functional DNA repair studies were performed and could not rule out xeroderma pigmentosum (XP), although they excluded XP-variant. Genotyping showed homozygosity of an ERCC4 pathogenic variant [NM_005236.2(ERCC4): c.1135C>T (p.Pro379Ser)], consistent with XP complementation group F (XP-F). XP-F is a rare form of XP and is significantly underdiagnosed due to its often milder clinical manifestations. Our case is the fourth case of XP-F that is reported in the UK, and shares similar characteristics with the other three published cases (Fassihi H, Sethi M, Fawcett H et al. Deep phenotyping of 89 xeroderma pigmentosum patients reveals unexpected heterogeneity dependent on the precise molecular defect. *Proc Natl Acad Sci U S A* 2016; 113: E1236–45). Notably, the identified pathogenic variant has an allele frequency of 0.3%, suggesting that around 10 individuals per million could be homozygous for this variant, equivalent to roughly 600 cases of XP-F currently in the UK. This case raises the importance of not underestimating ‘sunburn’ – attending accident and emergency with severe episodes following relatively minor sun exposure should be a red flag, requiring consideration of DNA repair diseases, even in the absence of any other features of XP. Moreover, it underscores the need to refer patients with abnormal photosensitivity to a specialist photodiagnostics service for comprehensive assessment and, where required, to the national XP service. It is likely that there is a significant number of ‘easy sunburners’, who are not currently recognized as having XP-F due to underdiagnosis, so we wish to raise awareness of this important condition.