**ABSTRACT**

**BACKGROUND**: Patients with genodermatosis such as Gorlin syndrome (GS) and Xeroderma pigmentosum (XP) require a close follow-up for early diagnosis and treatment of skin cancer. We aimed to evaluate the efficacy of methyl-aminolevulinate (MAL) photodynamic therapy (PDT) in basal cell carcinomas (BCCs) from patients with GS and XP, and to determine the utility of reflectance confocal microscopy (RCM) in the diagnosis and the evaluation of therapeutic response.

**PATIENTS AND METHODS**: We included four patients with GS and two siblings with XP. Single or multiple lesions in localized areas were treated with 1-3 cycles of MAL PDT. RCM was performed before and 3 months after the treatment in target lesions in all the patients. Patients were followed up for 3 years.

**RESULTS**: In XP patients, we treated 13 pigmented BCCs on the face. All the lesions responded to the treatment and six lesions showed a complete clinical clearing. In GS patients, facial or trunk areas with multiple BCCs were treated (up to 200). Complete clinical remission was obtained in 25-67% of the lesions. Some nodular and pigmented lesions failed to achieve a complete remission. RCM could identify already described confocal features for BCC. Tumour remissions could be assessed by this technique.

**CONCLUSIONS**: Methyl-aminolevulinate PDT may be useful for the treatment of superficial BCC in GS and XP. In some nodular lesions, PDT may complement surgery reducing tumour size. RCM may be regarded in the future as a complementary technique in BCC for the diagnosis and post-treatment assessment to non-invasive therapeutic modalities.