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Precision Medicine Approach in Advanced Squamous Cell Carcinoma

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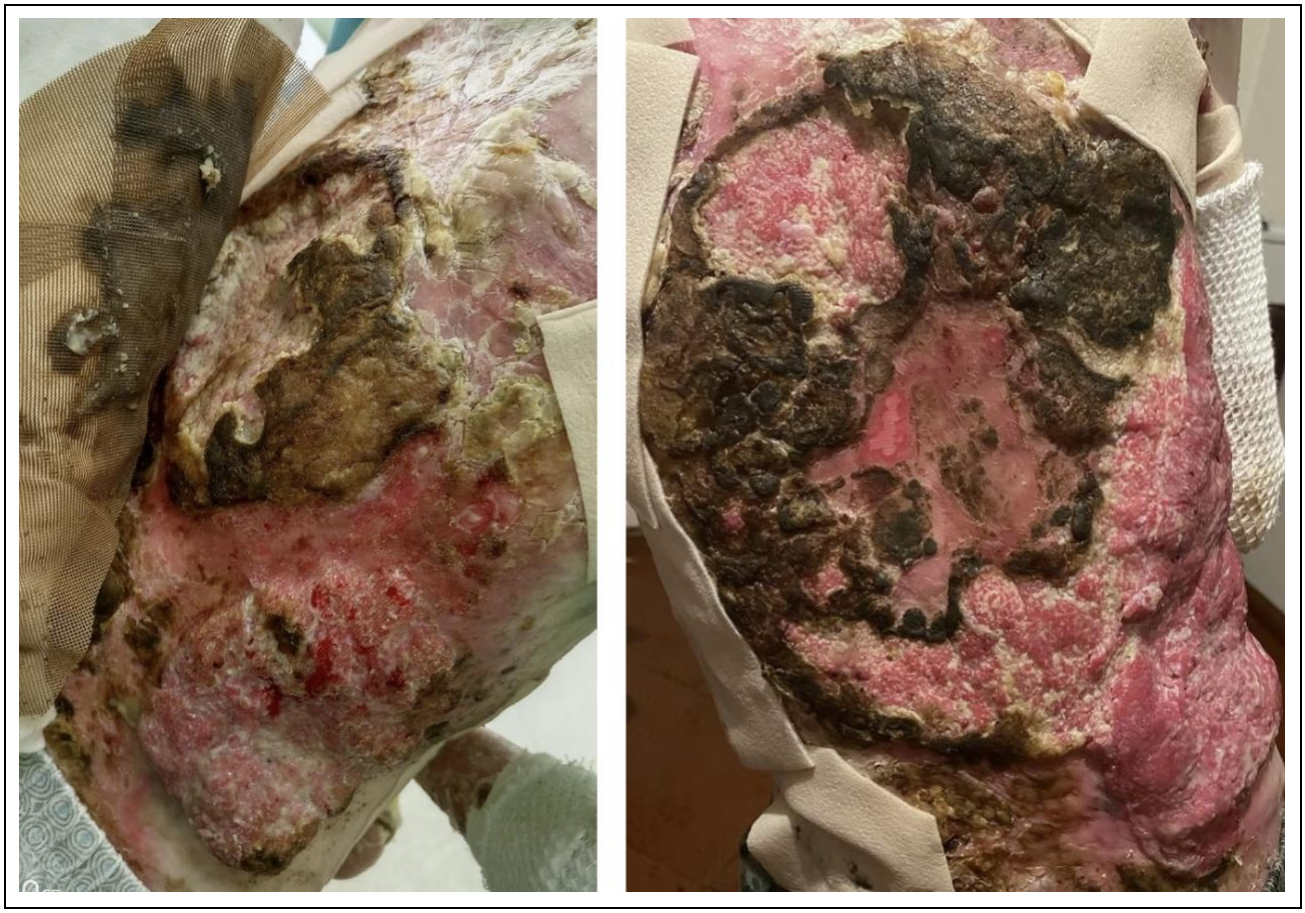

Clinical Image

A 31-year-old male with recessive Dystrophic Epidermolysis Bullosa (DEB) presented with two large, ulcerated lesions on his back, exhibiting dimensional increase and pain. Biopsy confirmed the diagnosis of Squamous Cell Carcinoma, commonly associated with DEB. Staging revealed no evidence of metastatic disease.

Given the lack of feasibility for surgical resection or radiotherapy, first-line treatment with Cemiplimab 350 mg tri-weekly was initiated. Between the 6th and 10th cycles, patient exhibited tumour progression with increasing pain. Next-Generation Sequencing was performed and an ATMR337C gene mutation was identified supporting the introduction of Niraparib at 100 mg/day. The patient remained on Niraparib for approximately one year, experiencing clinical benefit with pain control and maintenance of some autonomy. However, disease progression led to the patient's death in February 2024. This case underscores the potential of precision medicine to offer additional lines of treatment beyond standard options in rare genetic disorders with high malignant potential.