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Management Challenges in Gorlin Syndrome: A Case Report and Therapeutic Review

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Introduction

Gorlin syndrome, also known as basal cell nevus syndrome, is a rare autosomal dominant disorder characterized by multiple basal cell carcinomas (BCCs), odontogenic keratocysts, and skeletal abnormalities. It results from mutations in the PTCH1 gene, leading to dysregulation of the Sonic Hedgehog (SHH) signaling pathway. The management of patients with Gorlin syndrome is complex due to the high tumor burden, risk of recurrence, and potential for systemic involvement. We report a representative case and discuss the evolving therapeutic landscape for managing multiple BCCs in this condition.

Materials and Methods

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Results

A 65-year-old male patient, born from a consanguineous marriage, was referred to the dermatology department for management of multiple cutaneous tumors. His medical history included 32 sessions of radiotherapy at age 36 for skin tumors and regular follow-up for surgically treated multiple cutaneous lesions. Clinical examination revealed numerous basal cell carcinomas, palmar and plantar pits, and odontogenic keratocysts, confirming the diagnosis of Gorlin syndrome. A subsequent CT scan of the face and brain confirmed the presence of the suspected jaw cysts and showed no cerebral abnormalities. No evident bone abnormalities or neurological deficits were noted. The patient was referred to explore alternative therapeutic options due to the high number of tumors and the limitations of repeated surgical excisions.

Conclusions

Our case illustrates the central therapeutic challenge in Gorlin syndrome: controlling a high, recurrent tumor burden while minimizing cumulative morbidity. While surgical excision remains a cornerstone for definitive treatment, its iterative use can lead to significant functional and aesthetic compromise, as seen in this patient's decades-long history. Alternative strategies must therefore be integrated. Photodynamic therapy is a valuable non-invasive option for superficial BCCs, offering excellent cosmesis. For more advanced or numerous lesions, targeted SHH pathway inhibitors like vismodegib and sonidegib represent a paradigm shift, yet their long-term use is limited by cost, side effects, and acquired resistance, making them more suitable for advanced rather than lifelong prophylactic management. Other modalities, such as topical imiquimod or itraconazole, play ancillary roles. Crucially, radiotherapy is generally contraindicated due to its tumorigenic potential, a principle underscored by this patient's history. In summary, no single therapy is ideal for Gorlin syndrome. Effective management necessitates a personalized, multidisciplinary approach that strategically combines surgical, topical, and systemic modalities, adapting to disease progression and patient tolerance. This case reinforces the need for continued research into more sustainable targeted therapies and optimized treatment protocols to improve long-term quality of life for affected individuals.