RESEARCH PAPER

A Rare Case of Gorlin-Goltz Syndrome with Multiple Basal Cell Carcinoma Lesions: A Multidisciplinary Surgical Approach

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ABSTRACT

Background: Gorlin-Goltz Syndrome, also known as nevoid basal cell carcinoma syndrome (NBCCS), is a rare autosomal dominant disorder marked by multiple basal cell carcinomas (BCCs), odontogenic keratocysts, and systemic skeletal anomalies. Early diagnosis is crucial due to the syndrome's potential for multisystem involvement and malignancy risk. This case aims to highlight the clinical spectrum, diagnostic evaluation, and surgical management strategies in a patient with recurrent BCCs in the context of Gorlin syndrome, while emphasizing the role of multidisciplinary care in optimizing outcomes.

Clinical Presentation: A case of a 40-year-old Indian male presenting with multiple cutaneous lesions, including ulcerated pigmented nodules, predominantly on the scalp, limbs, and torso. He had a prior history of basal cell carcinoma excisions and a superficial parotidectomy with neck dissection. Systemic evaluation revealed hallmark features of Gorlin syndrome: macrocephaly, frontal bossing, odontogenic keratocysts (on orthopantomogram), and calcification of the falx cerebri (on CECT brain). Wide local excision (WLE) was performed for a symptomatic ulcerated lesion on the left hand. Histopathology confirmed recurrence of BCC. No systemic metastasis or skeletal anomalies were noted, though mild pulmonary arterial hypertension was detected on echocardiography.

Conclusion: The case highlights the importance of early recognition and comprehensive management in Gorlin-Goltz syndrome. Multidisciplinary surgical intervention, supported by regular follow-up and systemic evaluation, is essential to prevent complications and recurrence. The case adds to the scarce literature on Indian presentations of Gorlin syndrome and reinforces the need for genetic counselling and vigilant long-term care.

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INTRODUCTION

Gorlin-Goltz Syndrome, also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS), is a rare autosomal dominant genodermatosis caused by mutations in the PTCH1 gene located on chromosome 9q22.3 (1). It is characterized by a classical triad: multiple basal cell carcinomas (BCCs), odontogenic keratocysts (OKCs) of the jaw, and skeletal anomalies, often including bifid ribs, frontal bossing, and macrocephaly (2). The syndrome was first described by Gorlin and Goltz in 1960 and presents a wide spectrum of clinical manifestations that may affect the skin, skeleton, eyes, central nervous system, and other organs. Early detection is essential due to the potential for malignancy and cumulative morbidity from multiple tumors, especially BCCs (3).

Though common in Caucasian populations, Gorlin-Goltz Syndrome is rarely reported in the Indian demographic, and clinical recognition can be challenging in the absence of a strong family history or delayed presentation (4). Diagnosis is usually clinical but may be supported by genetic testing and imaging, particularly when evaluating for associated skeletal deformities or intracranial calcifications (5).

Management involves a multidisciplinary team including dermatologists, oncologic surgeons, maxillofacial specialists, radiologists, and genetic counselors. The cornerstone of treatment is surgical excision of tumors and cysts, supplemented by routine surveillance for new lesions. Avoidance of ionizing radiation is critical in these patients due to their increased sensitivity.

We present the case of a 40-year-old Indian male with a long-standing history of cutaneous lesions and multiple

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surgeries who was diagnosed with Gorlin-Goltz Syndrome. The case exemplifies the importance of coordinated multidisciplinary intervention in managing recurrent BCCs and associated manifestations. This report not only expands the clinical understanding of NBCCS in non-Western populations but also reiterates the need for lifelong surveillance and systemic evaluation to prevent recurrence and morbidity.

CASE PRESENTATION

Patient Profile

A 40-year-old male, reported to the Surgical Oncology department at KLES Dr. Prabhakar Kore Hospital and MRC, Belagavi, with complaints of multiple pigmented lesions over his scalp, face, chest, upper and lower limbs. A particularly ulcerated lesion over the left hand that had recently increased in size prompted his hospital visit. The patient had undergone prior excision of scalp lesions diagnosed as basal cell carcinoma (BCC), and a superficial parotidectomy with selective neck dissection in 2017. There was no relevant family history of similar conditions or malignancies.

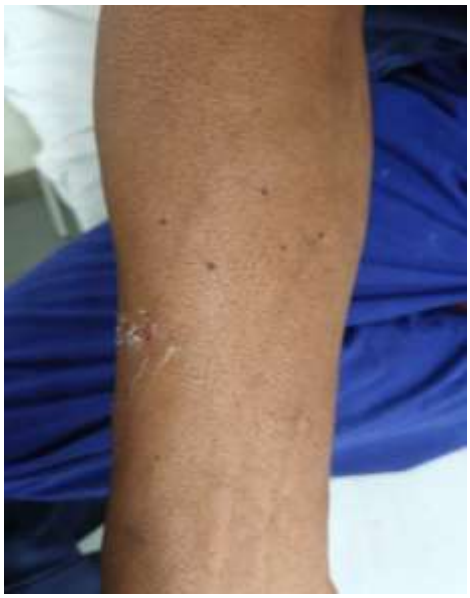

Figure 1: Upper limb image showing multiple pigmentated lesions

On general physical examination, the patient was conscious, oriented, and afebrile. His vital parameters were within normal limits. There were no signs of pallor, icterus, clubbing, lymphadenopathy, or pedal edema. The head circumference was measured at 61 cm, suggestive of macrocephaly. Distinct craniofacial abnormalities, including frontal bossing, a broad nasal root, and mild mandibular prognathism, were observed and are illustrated in Figure 2. These features are consistent with the phenotypic manifestations of Gorlin-Goltz Syndrome.

Figure 2: Frontal clinical image showing macrocephaly and mild frontal bossing in the patient.

On cutaneous examination, multiple hyperpigmented macules, papules, and nodular lesions were observed over various body regions. The face and scalp exhibited several lesions, some of which were ulcerated with overlying crusting. Similar dome-shaped lesions, resembling basal cell nevi, were also present on the forearms and legs. basal cell carcinomas, consistent with the clinical profile of Gorlin-Goltz Syndrome.

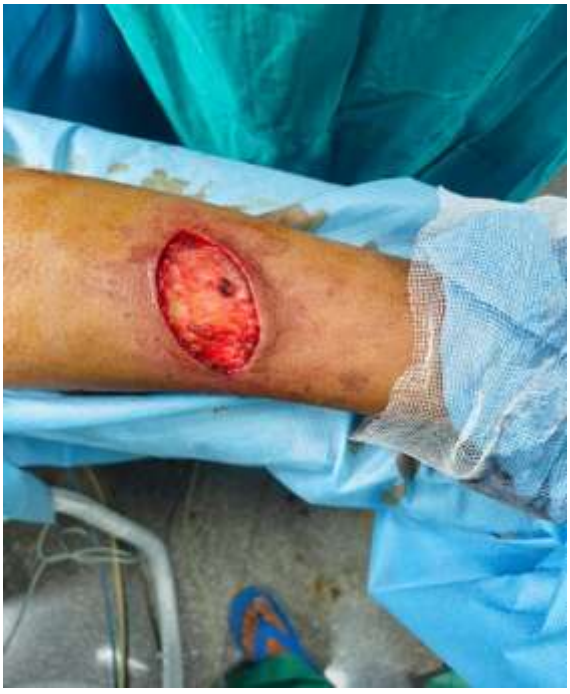

Figure 3: Post-operative image showing complete resection of Basal Cell Carcinoma.

Panoramic radiograph (Orthopantomogram – OPG) revealed multiple bilateral mandibular radiolucent lesions, consistent with odontogenic keratocysts (OKCs)

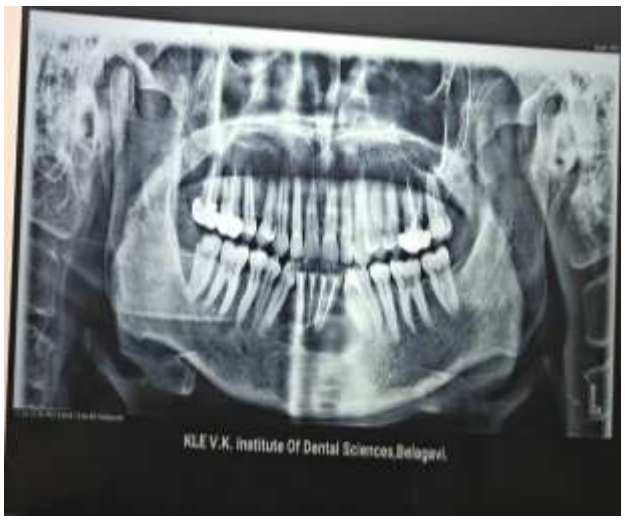

Figure 4: OPG showing multiple radiolucent cysts in the mandible, consistent with odontogenic cysts.

Neurological Evaluation

Contrast-enhanced CT (CECT) of the brain showed linear calcifications along the falx cerebri, which is a major diagnostic criterion in Gorlin-Goltz Syndrome

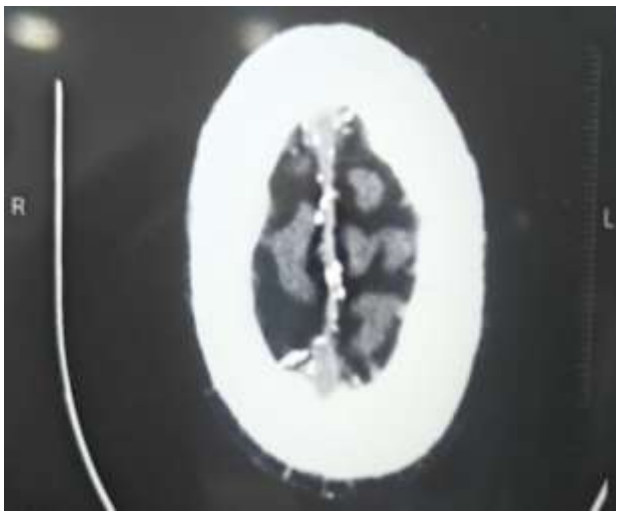

Figure 5: CECT of brain showing linear calcification along the falx cerebri

Cardiovascular and Respiratory Evaluation

Electrocardiogram (ECG) findings showed sinus bradycardia. Two-dimensional echocardiography demonstrated trivial mitral regurgitation and mild pulmonary arterial hypertension, with preserved left ventricular function. Computed tomography (CT) of the thorax revealed no evidence of pulmonary nodules or skeletal abnormalities.

Laboratory Investigations

Haematological and serological investigations were within normal limits. Hemoglobin was 13.8 g/dL, and the total white blood cell count was 4,600/cmm. The platelet count measured 2.04 lakh/cmm. Differential leukocyte count revealed 64% neutrophils, 23% lymphocytes, 8% monocytes, and 5% eosinophils, all within physiological ranges. The international normalized ratio (INR) was 1.00, indicating normal coagulation status. Serological tests for HIV, HBsAg, and HCV were non-reactive.

Provisional Diagnosis

Based on clinical and radiological findings, the patient fulfilled at least three major diagnostic criteria for Gorlin-Goltz Syndrome: multiple basal cell carcinomas, odontogenic keratocysts, calcification of the falx cerebri, and macrocephaly.

Treatment Plan

The patient was planned for wide local excision (WLE) of the ulcerated lesion over the left hand under monitored anaesthesia care. Intraoperatively, the lesion was excised with a 1 cm margin and sent for histopathological examination, with the report awaited. The postoperative recovery was uneventful. Medications prescribed included Augmentin, Pantodac, Emset, Enzoflam, and Chymoral Forte. Suture removal was scheduled on postoperative day 12. The patient was advised to continue regular follow-up with dermatology and dental departments.

DISCUSSION

Gorlin-Goltz Syndrome (GGS), or Nevoid Basal Cell Carcinoma Syndrome (NBCCS), is a rare hereditary condition with multisystem involvement, most notably presenting with early-onset and multiple basal cell carcinomas (BCCs) (6). The syndrome has an autosomal dominant pattern of inheritance, typically involving a mutation in the PTCH1 tumor suppressor gene, which encodes a receptor in the hedgehog signalling pathway (7). The hallmark of this condition is the development of numerous BCCs in sun-exposed areas, often beginning in adolescence or early adulthood, although this patient presented in his fourth decade of life with recurrent lesions and previous surgical history (8).

This case is particularly significant due to the presence of multiple diagnostic criteria, including multiple basal cell carcinomas (BCCs), odontogenic cysts of the mandible, calcification of the falx cerebri, macrocephaly with frontal

bossing, and characteristic skin lesions along with skeletal deformities (3,9).

The patient's head circumference measured over 61 cm, consistent with macrocephaly, one of the major diagnostic criteria of GGS. Moreover, CECT brain revealed calcifications of the falx cerebri, which is another classic radiological finding in this syndrome. Odontogenic keratocysts were identified via orthopantomogram (OPG), consistent with jaw cysts seen in nearly 90% of affected individuals.

The cutaneous findings, including multiple pigmented nevi and ulcerated lesions on the extremities, especially the left hand, aligned with typical BCC morphology. The patient had previously undergone excision of BCC on the scalp and superficial parotidectomy with selective neck dissection highlighting the recurrent nature of lesions and the progressive burden of disease.

What makes this case especially unique in the Indian context is the underreporting and underdiagnosis of Gorlin Syndrome, particularly in patients without a positive family history. Many individuals remain undiagnosed or misdiagnosed for years until classical skeletal and dermatologic features appear.

Systemic investigations revealed no pulmonary or skeletal metastases. ECG showed sinus bradycardia, while echocardiography indicated mild pulmonary arterial hypertension (PAH), although the exact correlation of cardiac abnormalities with GGS is not well established in literature and requires further evaluation.

Management of Gorlin Syndrome is challenging because of the recurrent and multiplicative nature of basal cell carcinomas (BCCs). Wide local excision (WLE) remains the standard approach for isolated and accessible lesions. However, due to the high risk of recurrence and the development of multiple lesions over time, alternative or adjunctive treatment modalities may be required. These include topical 5-fluorouracil or imiquimod for early or superficial lesions, photodynamic therapy for selective lesion control, and systemic therapy with hedgehog pathway inhibitors such as vismodegib or sonidegib in advanced or refractory cases.

Another critical aspect of management is avoiding radiotherapy, as patients with PTCH1 mutations have heightened radiosensitivity, increasing the risk of radiation-induced malignancies.

Psychological and cosmetic burden is considerable in these patients. The extensive cutaneous involvement, repeated surgeries, and disfigurement especially in visible areas like the face and scalp warrant psychosocial support and dermatological counselling.

In this patient, a multidisciplinary approach was essential. The surgical oncologist led the excision of cutaneous lesions, while the dental team identified jaw cysts and planned future maxillofacial management. Radiology contributed by evaluating both skeletal and neurological anomalies. This highlights the need for lifelong follow-up, genetic counselling, and screening of first-degree relatives. Genetic testing for PTCH1 mutation, although not performed in this case, is recommended where resources permit.

This case underscores the need for clinical vigilance in detecting Gorlin Syndrome in Indian patients, especially those presenting with early or multiple BCCs, odontogenic cysts, or craniofacial anomalies. Raising awareness among dermatologists, dentists, and primary care physicians is vital to enable early referral and prevent long-term morbidity.

CONCLUSION

This case highlights the importance of early recognition and multidisciplinary management in Gorlin-Goltz Syndrome. The presence of multiple BCCs, odontogenic keratocysts, and falx cerebri calcification confirms the diagnosis. Surgical excision remains effective, but long-term follow-up and systemic evaluation are essential due to the syndrome's recurrent nature. Greater awareness and early diagnosis can help reduce complications and improve quality of life in affected individuals.

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