International Journal of Research in Dermatology
Bhattacharya S. Int J Res Dermatol. 2026 Jul;12(4):351-354
https://www.ijord.com

DOI: https://dx.doi.org/10.18203/issn.2455-4529.IntJResDermatol20261936

Case Report

Gorlin-Goltz syndrome presenting with multiple basal cell carcinomas and odontogenic keratocysts: a case report

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Received: 23 February 2026
Revised: 06 April 2026
Accepted: 05 April 2026

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ABSTRACT

Gorlin-Goltz syndrome, also known as nevoid basal cell carcinoma syndrome (NBCCS), is a rare autosomal dominant disorder characterized by multiple basal cell carcinomas, odontogenic keratocysts, skeletal anomalies, and intracranial calcifications. It results from mutations in the PTCH1 gene, leading to dysregulation of the Hedgehog signaling pathway. Early diagnosis is crucial due to its multisystem involvement and potential for malignancy. We report a case of a 45-year-old male presenting with multiple facial lesions of long duration associated with jaw swelling and palmar pits. Radiological evaluation including CBCT revealed multiple well-defined radiolucencies involving the maxilla and mandible along with bifid ribs and calcification of the falx cerebri. Histopathological examination confirmed basal cell carcinoma and odontogenic keratocysts. Based on clinical, radiological, and histopathological findings, a diagnosis of Gorlin-Goltz syndrome was established. The patient was managed with a multidisciplinary approach including dermatological and surgical interventions with satisfactory outcome on follow-up. This case highlights the importance of early recognition, multidisciplinary management, and long-term surveillance in preventing complications associated with this syndrome.

Keywords: Gorlin syndrome, NBCCS, Basal cell carcinoma, Odontogenic keratocyst, CBCT

INTRODUCTION

Gorlin-Goltz syndrome, also known as NBCCS, is a rare multisystem genetic disorder characterized by developmental abnormalities and predisposition to neoplasms.[1] It was first described by Jarisch and White in 1894 and later delineated by Gorlin et al.[2]

The syndrome follows an autosomal dominant inheritance pattern with high penetrance and variable expressivity. It is caused by mutations in the PTCH1 gene located on chromosome 9q22.3-q31, resulting in abnormal activation of the Sonic Hedgehog signaling pathway.[3]

The prevalence ranges from 1 in 57,000 to 1 in 256,000 individuals with no gender predilection.[4] Clinical manifestations typically appear during adolescence but may present later in life.

Major diagnostic criteria include multiple basal cell carcinomas, odontogenic keratocysts, palmar or plantar pits, calcification of the falx cerebri, and bifid ribs.[5] Early diagnosis is essential for surveillance and prevention of complications such as aggressive basal cell carcinomas and medulloblastoma.[6]

CASE REPORT

A 45-year-old male presented with multiple lesions over the face for the past 10 years. The lesions were initially small and gradually increased in size, ranging from 2×2 cm to 5×5 cm. Some lesions showed rolled borders with central ulceration.

On dermatological examination, multiple well-defined plaques with pearly margins and central indentation were observed over the face, clinically suggestive of basal cell carcinoma. Bilateral palmar pits (>3 in number) were noted.

The patient also complained of jaw swelling associated with pain, pus discharge, loosening of teeth, and trismus. There was no significant family history.

Systemic examination including cardiovascular, respiratory, and genitourinary systems revealed no abnormalities.

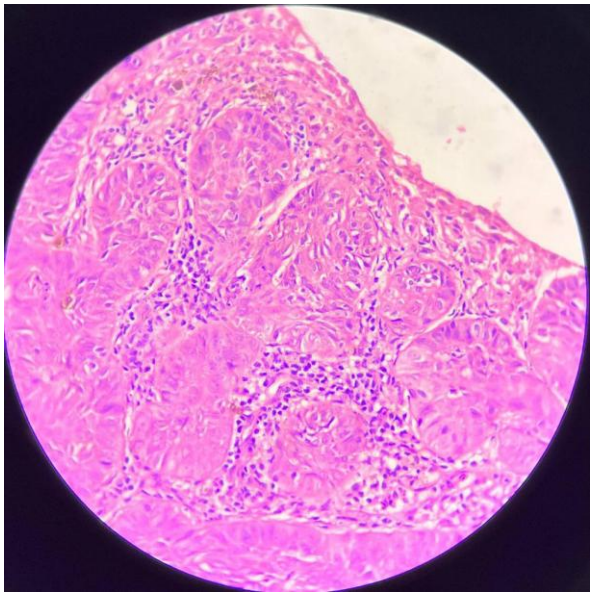

Figure 1: Histopathological findings (H&E stain, intermediate power).

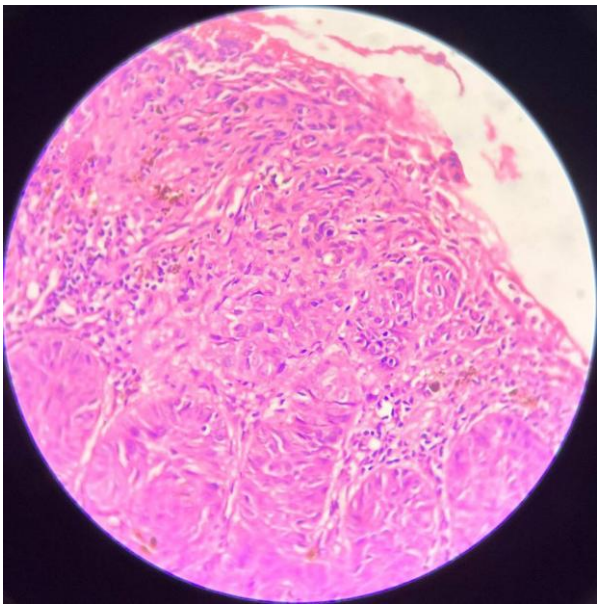

Figure 2: Histopathological findings (H&E stain).

Radiological findings

Cone beam computed tomography (CBCT) revealed multiple well-defined radiolucent lesions involving the right maxillary region, mandibular ramus, and symphyseal region. These findings were consistent with odontogenic keratocysts.

Chest imaging demonstrated bifid ribs, while CT scan of the skull showed bilamellar calcification of the falx cerebri.

Histopathological findings

Histopathological examination of the facial lesion revealed nests and islands of basaloid cells arising from the epidermis and extending into the dermis. Peripheral palisading, stromal retraction spaces, and increased mitotic activity were noted, consistent with basal cell carcinoma.

The histopathology report from Dr. R. Ahmed Dental College confirmed features of odontogenic keratocyst. Sections showed odontogenic cystic lining supported by connective tissue stroma, with stratified epithelium exhibiting surface keratinization and palisaded basal cells. The underlying stroma was fibrocollagenous with scant inflammatory infiltrate.

Based on the presence of multiple basal cell carcinomas, odontogenic keratocysts, palmar pits, bifid ribs, and falx cerebri calcification, the diagnosis of Gorlin-Goltz syndrome was established.[7]

Figure 3: Clinical presentation of a well-demarcated, hyperkeratotic, and crusted plaque on the left malar region of the face.

Figure 4: Anterior view demonstrating the primary left zygomatico-malar skin lesion.

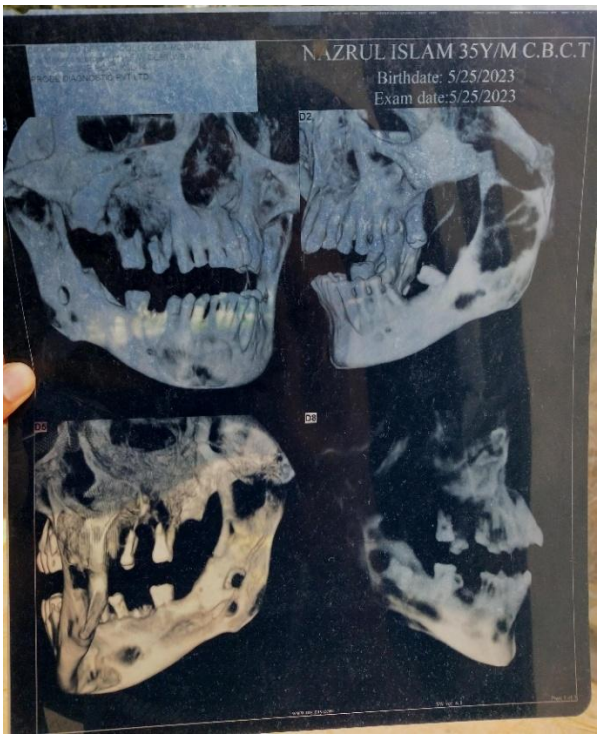

Figure 5: Radiological evaluation.

Treatment and follow-up

The patient was managed with a multidisciplinary approach involving dermatology and maxillofacial surgery teams.

Cutaneous lesions were treated using a combination of: Topical imiquimod 5% cream, surgical excision of larger lesions, cryotherapy for selected lesions.

Odontogenic keratocysts were managed by surgical enucleation and curettage.

The patient was advised strict photoprotection, including avoidance of ultraviolet exposure and regular use of sunscreen.

At 6-month follow-up, there was significant clinical improvement with no evidence of recurrence of cutaneous lesions or progression of jaw lesions. The patient remains under regular surveillance. Genetic counselling was advised.

DISCUSSION

Gorlin-Goltz syndrome is a rare multisystem disorder with variable clinical presentation. Diagnosis is based on established criteria requiring either two major criteria or one major and two minor criteria.[8]

In the present case, multiple major criteria including basal cell carcinomas, odontogenic keratocysts, palmar pits, bifid ribs, and falx cerebri calcification were present, confirming the diagnosis.

Basal cell carcinomas in NBCCS often present at an earlier age and tend to be multiple.[9] They may mimic benign lesions such as nevi or seborrheic keratosis, leading to delayed diagnosis.

Odontogenic keratocysts are among the earliest manifestations and may present with jaw swelling, pain, and dental abnormalities.[10] CBCT plays a crucial role in identifying these lesions and assessing their extent.

The absence of family history in this patient suggests a possible de novo mutation, which has been reported in a significant number of cases.[11]

Management of NBCCS requires a multidisciplinary approach. Treatment options for basal cell carcinoma include surgical excision, topical therapy, cryotherapy, and photodynamic therapy.[12]

Recent advances include Hedgehog pathway inhibitors such as vismodegib and sonedegib, which are effective in advanced or inoperable cases.[13]

Regular follow-up is essential for early detection of complications such as medulloblastoma, particularly in younger patients.[14]

CONCLUSION

Gorlin–Goltz syndrome is a rare genetic disorder with significant clinical implications due to its multisystem

involvement and oncogenic potential. This case highlights the importance of early recognition of characteristic clinical and radiological features. A multidisciplinary approach and long-term follow-up are essential for optimal management. Early diagnosis and appropriate intervention can significantly reduce morbidity and improve patient outcomes.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Bhattacharya S. Gorlin-Goltz syndrome presenting with multiple basal cell carcinomas and odontogenic keratocysts: a case report. Int J Res Dermatol 2026;12:351-4.