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Case Report

Comprehensive Cone-Beam Computed Tomographic Findings in Gorlin–Goltz Syndrome: A Case Report

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Abstract

Gorlin–Goltz syndrome (GGS), sometimes called nevoid basal cell carcinoma syndrome, is a rare genetic condition inherited in an autosomal dominant manner, most often resulting from changes in the PTCH1 tumour suppressor gene. It affects multiple body systems, with common features including jaw cysts (odontogenic keratocysts), skeletal abnormalities, and calcifications within the brain. Here, we describe an unusual case of GGS in a 24-year-old woman who came to us with a painless swelling in her jaw. Cone-beam computed tomography (CBCT) scans revealed several jaw cysts and a distinctive calcification of the falx cerebri, which met key diagnostic criteria for the syndrome. This case highlights how valuable CBCT and the expertise of dental professionals are for the timely detection and comprehensive care of individuals with Gorlin–Goltz syndrome.

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KEYWORDS: Gorlin–Goltz syndrome; nevoid basal cell carcinoma syndrome; odontogenic keratocyst; cone-beam computed tomography; falx cerebri calcification.

INTRODUCTION

Gorlin–Goltz syndrome (GGS), also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS), is a rare inherited multisystem disorder characterised by variable clinical expressivity despite complete penetrance. Gorlin and Goltz first described the syndrome in 1960 as a triad comprising multiple basal cell carcinomas, odontogenic keratocysts of the jaws, and skeletal anomalies such as bifid ribs [1]. BCNS results from pathogenic mutations in the PATCHED (PTCH1) gene, a tumor suppressor gene that plays a crucial role in the regulation of the hedgehog signalling pathway, which is essential for normal embryonic development and tissue homeostasis [2]. Disruption of this pathway predisposes affected individuals to abnormal cellular proliferation and tumor formation. Although BCNS is frequently inherited, approximately one-third of cases arise from spontaneous, or de novo, genetic mutations in individuals with no prior family history of the disorder [3].

The global prevalence of BCNS has been estimated to range between 1 in 50,000 and 1 in 150,000 individuals, with considerable variation reported across different geographic regions and ethnic populations [3]. This wide range may be attributed to differences in genetic backgrounds, environmental influences, and diagnostic awareness. Furthermore, the diverse and often subtle clinical manifestations of BCNS may result in underdiagnosis or delayed identification, particularly in younger patients or in settings with limited access to specialised diagnostic resources. Gorlin and Goltz originally described the syndrome based on a characteristic clinical triad consisting of multiple basal cell carcinomas (BCCs), odontogenic keratocysts of the jaws, and congenital skeletal anomalies [4]. Since that initial description, numerous additional features have been recognised, including craniofacial abnormalities, neurological findings, and a variety of cutaneous manifestations. To facilitate accurate and consistent diagnosis, standardised diagnostic criteria were subsequently established, encompassing five major and eight minor criteria. A definitive diagnosis of BCNS is made when either two major criteria or one major criterion in combination with two minor criteria are present [1].

Imaging plays a central role in the diagnosis and long-term management of BCNS, particularly in the evaluation of maxillofacial involvement. Cone-beam computed tomography (CBCT) has emerged as a highly effective imaging modality for assessing the craniofacial and skeletal manifestations associated with Gorlin–Goltz syndrome. CBCT allows precise three-dimensional visualization of jaw cysts, cortical bone changes, and other osseous abnormalities, facilitating early detection and accurate characterization of lesions. In addition, the relatively low radiation dose associated with CBCT compared with the conventional computed tomography makes it especially suitable for patients with BCNS, who often require repeated imaging examinations over the course of their lifetime.

limited literature that shows cone-beam computed tomography features of this condition, the present article reports a detailed case of Gorlin–Goltz syndrome with special emphasis on CBCT findings. By documenting the characteristic radiographic features and their diagnostic relevance, this report seeks to enhance understanding of the role of CBCT in the early diagnosis and effective management of patients with BCNS.

CASE REPORT

A 24-year-old female presented to the Department of Oral Medicine and Radiology at Sri Aurobindo College of Dentistry with a complaint of a slowly enlarging, painless swelling in the lower jaw that had been present for several months. There was no associated history of trauma, discharge, or discomfort. The patient's medical and dental histories did not reveal any significant findings. General physical examination showed that the patient was well built and adequately nourished. Extraoral assessment revealed noticeable facial asymmetry along with coarse facial appearance, mandibular prognathism, frontal bossing, and hypertelorism. No enlargement of regional lymph nodes was observed. Intraoral evaluation demonstrated a firm, non-tender swelling in the posterior region of the mandible, leading to partial obliteration of the buccal vestibule. The overlying mucosa appeared intact and healthy. Mild expansion of the buccal cortical plate was evident on palpation, and there were no signs of infection or mobility of the adjacent teeth.

To further assess the extent and nature of the lesion, cone beam computed tomography (CBCT) was performed, which revealed the following findings

Radiographic Findings (CBCT)

CBCT examination revealed multiple odontogenic abnormalities involving both jaws.

Dentition

Multiple over-retained primary teeth (53, 54, 55, 63, 64, 65, 83) with impacted permanent teeth (13, 14, 15, 23) and congenitally missing teeth (12, 22, 33, 34, 35) were noted. Physiologic root resorption was seen in 64, 65, and 83.

Maxilla

Tooth 13 was vertically impacted with its crown at the apical third of 53 and the root apex in proximity to the nasal floor and maxillary sinus. A well-defined pericoronal hypodensity arising from the CEJ surrounded its crown, consistent with a dentigerous cyst. Apical dilaceration was noted. The crown lay approximately 11 mm from the occlusal plane at an angulation of ~60°.

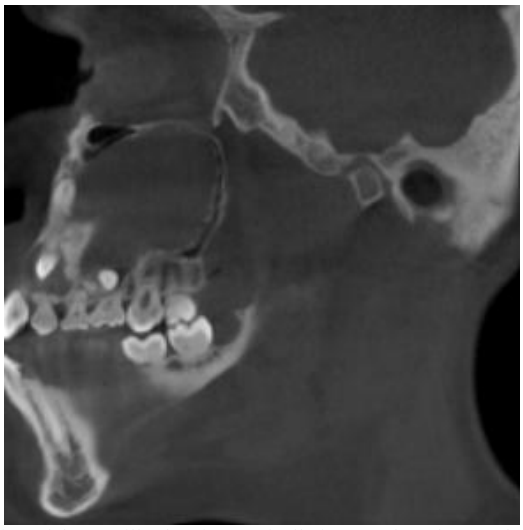


Figure 1: Sagittal CBCT section of the maxilla showing vertically impacted tooth 13 and an associated dentigerous cyst.

Tooth 14 was vertically impacted near the cervical third of 54, with apical dilaceration and an angulation of ~98°, while 15 was horizontally impacted, extending toward the middle third of 16.

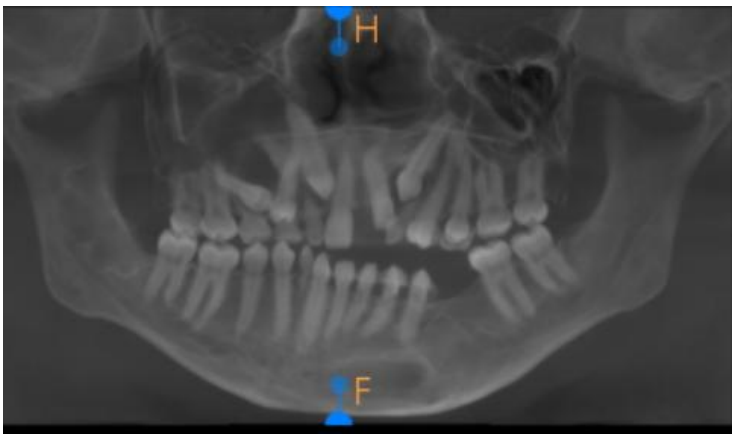


Figure 2: Sagittal CBCT section of the maxilla showing vertically impacted tooth 13 with an associated dentigerous cyst and root apex in proximity to the nasal floor and maxillary sinus.

A large well-defined, corticated, homogeneous isodense lesion occupied the right maxillary sinus, extending from the alveolar crest to the sinus roof and from the distal aspect of 14 to the posterior sinus wall. The lesion measured approximately 26.8 × 38.4 × 28.7 mm, caused thinning and expansion of the buccal and palatal cortical plates, and resulted in loss of visualization of the sinus floor lining.

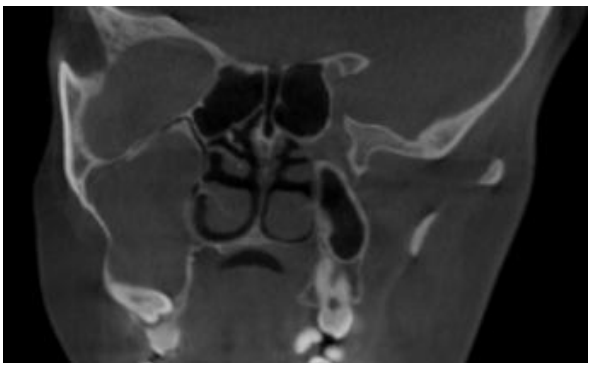


Figure 3: Sagittal CBCT section of the maxilla showing vertically impacted tooth 13 with its crown at the apical third of 53, an associated dentigerous cyst, and apical dilaceration

Mandible

A well-defined hypodense lesion with scalloped margins was present in the right posterior mandible, extending from distal to 47 to the ascending ramus. The lesion measured approximately 29.7 × 6.4 × 11.8 mm, showed homogeneous internal structure, caused lingual cortical expansion, and involved the inferior alveolar nerve canal.

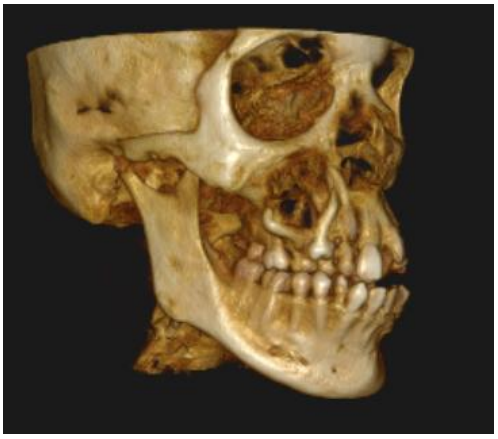


Figure 4: 3D images of maxillary and mandibular lesions

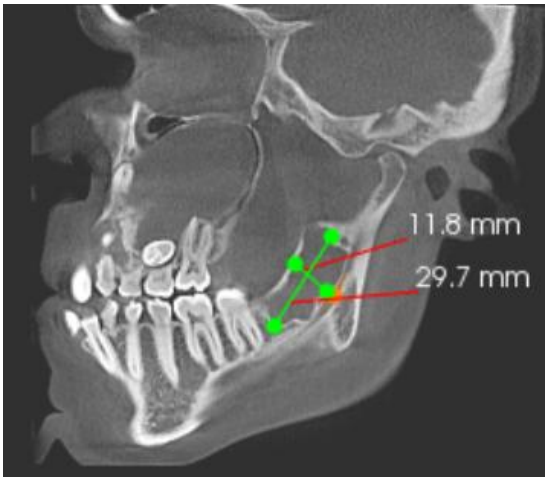
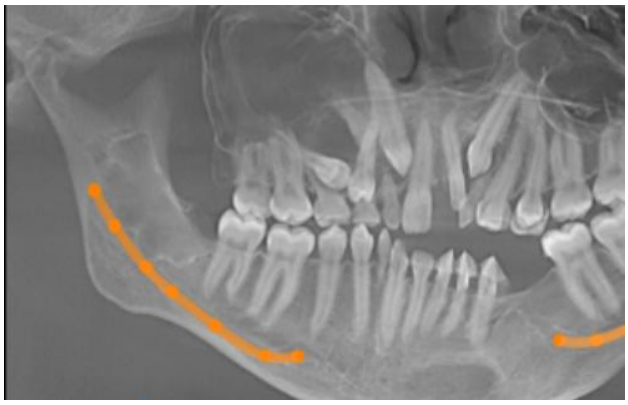


Figure 5: Dimensions of mandibular lesion



Most importantly, a linear, crescent-shaped intracranial calcification representing calcification of the falx cerebri was identified, which is a highly characteristic and diagnostically significant radiologic feature of Gorlin–Goltz syndrome.

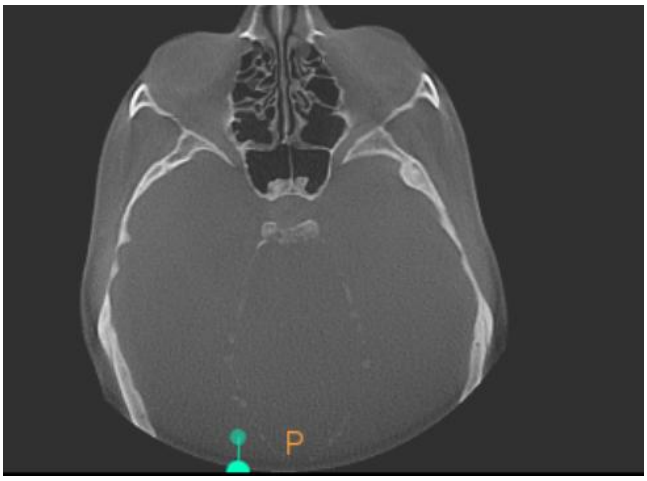


Figure 7: Axial CBCT section of the skull identifying a linear, crescent-shaped intracranial calcification of the falx cerebri.

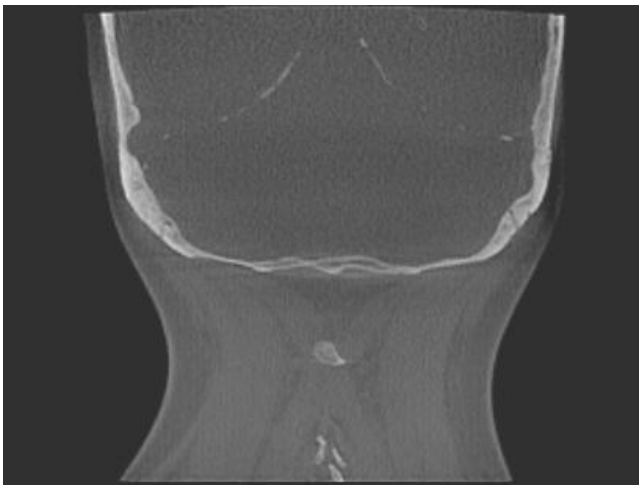


Figure 8: Coronal CBCT section of the skull identifying a linear, crescent-shaped intracranial calcification of the falx cerebri

Additional findings included the presence of multiple tonsilloliths, areas of idiopathic osteosclerosis in relation to teeth 13, 24, and 25, and remnants of predecessor primary teeth in the regions of 31 and 32; paranasal sinus variations such as left concha bullosa, aerated crista galli, and a Haller cell were also noted.

Gorlin–Goltz syndrome is characterized by multisystem involvement, with multiple odontogenic keratocysts (OKCs) representing a major diagnostic criterion. In the present case, CBCT revealed multifocal cystic lesions involving both the maxilla and mandible, displaying classic radiographic features of OKCs including well-defined corticated margins, homogeneous internal density, minimal buccolingual expansion, and involvement of the inferior alveolar nerve canal. The presence of falx cerebri calcification, another major diagnostic criterion, further strengthens the diagnosis. Additional findings such as multiple impacted teeth, dentigerous cyst formation, idiopathic osteosclerosis, and maxillary sinus involvement support the syndromic nature of the presentation. CBCT played a pivotal role in accurately delineating lesion extent, cortical involvement, neurovascular proximity, and associated developmental anomalies, underscoring its importance in early diagnosis, treatment planning, and long-term surveillance of patients with Gorlin–Goltz syndrome.

Table 1. Summary of Odontogenic Lesions Identified on CBCT

Right posterior maxilla	Well-defined corticated isodense lesion occupying maxillary sinus	26.8 × 38.4 × 28.7	Odontogenic keratocyst/ Dentigerous cyst
Right posterior mandible	Scalloped hypodense lesion involving IAN canal	29.7 × 6.4 × 11.8	Odontogenic keratocyst

Table 2. CBCT Findings Supporting Gorlin–Goltz Syndrome

Multiple OKCs	Maxilla and mandible	Major criterion
Falx cerebri calcification	Present	Major criterion
Impacted teeth	Multiple	Common associated finding
Dentigerous cyst	Tooth 13	Syndromic association

DISCUSSION

Gorlin–Goltz syndrome (GGS), also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS), is a rare inherited multisystem disorder characterized by variable clinical expressivity despite complete penetrance. Gorlin and Goltz first described the syndrome in 1960 as a triad comprising multiple basal cell carcinomas, odontogenic keratocysts of the jaws, and skeletal anomalies such as bifid ribs [1]. Subsequent studies expanded the phenotypic spectrum, leading to the establishment of standardized diagnostic criteria by Evans et al. and later refinement by Kimonis et al., which remain the most widely accepted framework for diagnosis [5]. According to these criteria, a definitive diagnosis is made when either two major criteria or one major criterion along with two minor criteria are fulfilled.

In the present case, diagnosis of Gorlin–Goltz syndrome was established based on the fulfillment of multiple major diagnostic criteria, supported by several associated findings. This diagnostic approach is consistent with previously published literature, where clinical and radiologic features have been emphasized as sufficient for diagnosis, particularly in the absence of genetic testing [5,10].

One of the most consistent and earliest manifestations of GGS is the presence of multiple odontogenic keratocysts (OKCs), which occur in up to 90% of affected individuals [6]. In the present case, CBCT revealed multiple well-defined cystic lesions involving both the maxilla and mandible, exhibiting characteristic features of OKCs such as corticated margins, homogeneous internal density, minimal buccolingual expansion, and involvement of the inferior alveolar nerve canal. Similar presentations have been reported by Varkhede et al., Lee et al., and Kapoor et al., who emphasized that multiple OKCs are often the first clinical indication of an underlying syndromic condition and may precede cutaneous manifestations by several years [7,8,11]. The early age of presentation and multiplicity of lesions in the present case closely parallels these observations.

Falx cerebri calcification, another major diagnostic criterion, was identified incidentally on CBCT in the present patient. Intracranial calcifications, particularly involving the falx cerebri, are among the most frequent radiologic findings in GGS, with prevalence increasing with age and reported in up to 85–90% of adult patients [9]. Similar findings have been highlighted by Kimonis et al. and Evans et al. as highly specific and diagnostically valuable, especially when detected during imaging performed for dental complaints [5,9]. The presence of falx cerebri calcification in the present case therefore significantly strengthens the diagnosis.

Basal cell carcinoma (BCC) is traditionally considered the hallmark feature of GGS; however, its absence does not preclude diagnosis. Several authors have documented delayed onset or complete absence of BCCs in younger patients and in individuals with darker skin pigmentation [6,10]. In the present case, no clinically evident BCCs were observed at the time of diagnosis. Similar findings were reported by Lee et al., who described a case of Gorlin–Goltz syndrome presenting with multiple OKCs in the absence of skin manifestations [8]. Indian case reports by Anchlia et al., Hegde and Shetty, and Nikam et al. have also documented patients lacking cutaneous involvement at presentation, supporting the observation that dermatologic features may be delayed in certain populations [6,12,13].

Dental developmental anomalies were prominent in the present case, including multiple impacted permanent teeth, over-retained deciduous teeth, congenitally missing teeth, and associated dentigerous cyst formation. Such findings have been well documented in GGS and are attributed to disruption of odontogenesis related to PTCH1 mutations [2]. Kapoor et al. emphasized that dental anomalies, particularly impacted teeth and delayed eruption, are common associated findings and may aid in early diagnosis in pediatric and young adult patients [11]. The dental findings in the present case are therefore consistent with previously reported patterns.

Advanced imaging played a crucial role in the present case. CBCT enabled precise three-dimensional assessment of lesion extent, cortical plate involvement, maxillary sinus occupation, and proximity to vital structures such as the inferior alveolar nerve. Similar advantages of CBCT have been highlighted in earlier studies, which emphasize its utility in early detection, surgical planning, and long-term surveillance of syndromic OKCs [7,8]. Given the high recurrence rate reported for GGS-associated OKCs, detailed baseline imaging is essential.

Neurological manifestations such as medulloblastoma and meningioma, although reported in the literature, occur at relatively low frequencies [6]. No such findings were identified in the present case, which is consistent with the majority of reported cases and does not detract from the diagnosis.

From an epidemiological perspective, Gorlin–Goltz syndrome is considered extremely rare in the Indian population. Unlike Western literature, where prevalence estimates range between 1 in 50,000 and 1 in 150,000 individuals, Indian data are largely confined to isolated case reports and small case series [3]. Reports by Anchlia et al., Hegde and Shetty, Nikam et al., and Kapoor et al. underscore the scarcity of documented cases and suggest that the condition is likely underdiagnosed due to lack of awareness and delayed recognition of non-cutaneous manifestations [6,11,12,13]. The present case adds to the limited Indian literature and highlights the importance of dental and radiologic evaluation in identifying this rare syndrome.

Overall, the present case satisfies two major diagnostic criteria—multiple odontogenic keratocysts and calcification of the falx cerebri—together with several associated dental and

maxillofacial abnormalities. These observations are consistent with findings reported in earlier studies and support the notion that Gorlin–Goltz syndrome can initially manifest predominantly through oral and radiographic features, even in the absence of cutaneous lesions.

CONCLUSION

Gorlin–Goltz syndrome, also known as nevoid basal cell carcinoma syndrome, represents a rare but clinically significant inherited disorder with wide phenotypic variability and multisystem involvement. The present case highlights the pivotal role of oral and maxillofacial manifestations—particularly multiple odontogenic keratocysts—in facilitating early recognition of this syndrome, often before the appearance of classical cutaneous features. The identification of multiple jaw cysts in association with falx cerebri calcification fulfilled the major diagnostic criteria, thereby establishing the diagnosis even in the absence of basal cell carcinomas.

Cone-beam computed tomography proved invaluable in the comprehensive evaluation of this patient by enabling precise delineation of lesion extent, cortical bone involvement, neurovascular proximity, maxillary sinus involvement, and associated developmental anomalies. Such detailed radiographic assessment is essential not only for accurate diagnosis but also for effective treatment planning and long-term surveillance, given the high recurrence rate of odontogenic keratocysts associated with Gorlin–Goltz syndrome.

This case reinforces existing literature indicating that Gorlin–Goltz syndrome may present predominantly with dental and radiologic findings, especially in younger individuals and in populations with darker skin pigmentation where cutaneous manifestations may be delayed or absent. The rarity of reported cases in the Indian population further underscores the likelihood of underdiagnosis and the need for heightened clinical awareness among dental professionals.

Early diagnosis through meticulous clinical examination and advanced imaging allows timely multidisciplinary management, appropriate genetic counselling, and lifelong follow-up, thereby reducing morbidity and improving patient outcomes. Dental practitioners, particularly oral radiologists and oral medicine specialists, play a crucial role in the early detection of this syndrome and should maintain a high index of suspicion when encountering multiple jaw cysts or associated craniofacial anomalies.

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