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Case Report

Odontogenic Cysts in Gorlin Goltz Syndrome - Clinicoradiological Presentation and Management

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Abstract

Gorlin–Goltz syndrome, or nevoid basal cell carcinoma syndrome, is an uncommon autosomal dominant disorder characterized by multiple odontogenic keratocysts, basal cell carcinomas, and a spectrum of skeletal and developmental anomalies. We present the case of a 13-year-old male who reported with progressive mandibular swellings and facial asymmetry. Clinical examination and panoramic radiography revealed multiple well-circumscribed radiolucent lesions involving both jaws, consistent with odontogenic keratocysts. The diagnosis was established based on clinic-radiological findings and confirmed histo-pathologically. Surgical enucleation of the cysts was performed with adjunctive measures to reduce the risk of recurrence. This case underscores the importance of early recognition of Gorlin–Goltz syndrome in paediatric patients, the necessity of multidisciplinary management, and the imperative for long term surveillance to mitigate morbidity and detect associated systemic manifestations.

Introduction

Gorlin–Goltz syndrome, also referred to as nevoid basal cell carcinoma syndrome (NBCCS), is a rare multisystemic genetic disorder inherited as an autosomal dominant genetic disorder. It predisposes affected individuals to a wide spectrum of neoplasms and developmental anomalies. The classical triad of multiple basal cell carcinomas (BCCs), odontogenic keratocysts (OKCs) of the jaws, and bifid ribs was first described by Gorlin and Goltz in 1960 [1]. Since then, additional features such as intracranial calcifications, skeletal abnormalities, and craniofacial dysmorphism have been recognized, highlighting the broad clinical spectrum of this condition.

The odontogenic keratocyst was initially reported by Philipsen in 1956 [2]. Due to its aggressive nature, infiltrative growth, and high recurrence rate, the World Health Organization (WHO) reclassified the lesion in 2005 as a keratocystic odontogenic tumour, reflecting its neoplastic potential. In the revised 2017 WHO classification, however, it was redefined once again as a cyst, underscoring the ongoing debate regarding its biological behaviour [3]. Despite this reclassification, OKCs remain clinically significant because they tend to recur and are strongly associated with Gorlin–Goltz syndrome.

Approximately 75% of patients with Gorlin–Goltz syndrome present with multiple OKCs, making them one of the most consistent and distinctive diagnostic features of the disorder [4]. Radiographically, these lesions typically appear as multiple well-defined radiolucencies within the jaws, often associated with cortical expansion and displacement of teeth. Thomas introduced the term polycystomya to describe this characteristic radiographic presentation of multiple cystic lesions. The presence of such findings, particularly in young patients, should raise clinical suspicion for Gorlin–Goltz syndrome.

Given the multisystemic nature of the disorder, early recognition and accurate diagnosis are essential for effective management. A multidisciplinary approach involving oral and maxillofacial surgeons, dermatologists, radiologists, and genetic counsellors is often required to address the diverse manifestations and reduce morbidity. This case report discusses the clinic-radiological features, diagnostic considerations, and management of multiple OKCs in a 13-year-old male patient with Gorlin–Goltz syndrome, emphasizing the importance of comprehensive evaluation and long-term follow-up.

Case Report

A 13-year-old male presented to the Department of Oral and Maxillofacial Surgery at Panineeya Institute of Dental Sciences, Hyderabad, with swelling on the right side of his face for one month. The patient had a history of an odontogenic keratocyst treated five years earlier with enucleation, curettage, and Carnoy's solution. Clinical examination revealed facial asymmetry with swelling over the right middle and lower thirds of the face and frontal bossing as shown in (Figure 1). Intraoral examination of the maxilla showed diffuse swelling obliterating the vestibule from 15 to 21, with missing 13 and 11 and retained 55 and 63. Intraoral examination of the mandible revealed swelling from 31 to 45, with missing 34 and 43, and retained 72 and 83.

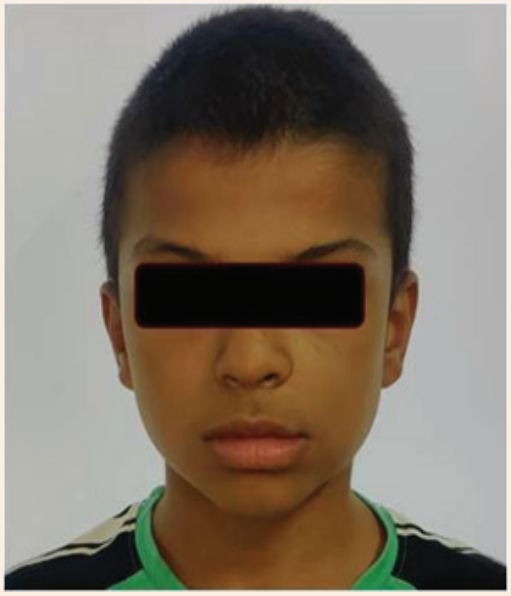

Figure 1: Frontal asymmetry of the face.

Radiological investigations included an orthopantomogram (OPG), which demonstrated multiple radiolucency's in both maxilla and mandible (Figure 2). Cone beam computed tomography (CBCT) of the maxilla revealed buccal cortical thinning and displacement of the sinus floor by an impacted 13, (Figure 3) while CBCT of the mandible showed a multilocular radiolucency extending from 33 to 46 with cortical expansion, displacement of the inferior alveolar canal, and impacted 43 displaced inferiorly. Additional radiolucencies were noted around developing 38 and 48 (Figure 4). The chest radiograph did not reveal bifid ribs, but the lateral cephalogram confirmed frontal bossing (Figure 5), and the skull radiograph demonstrated calcification of the falx cerebri (Figure 6).

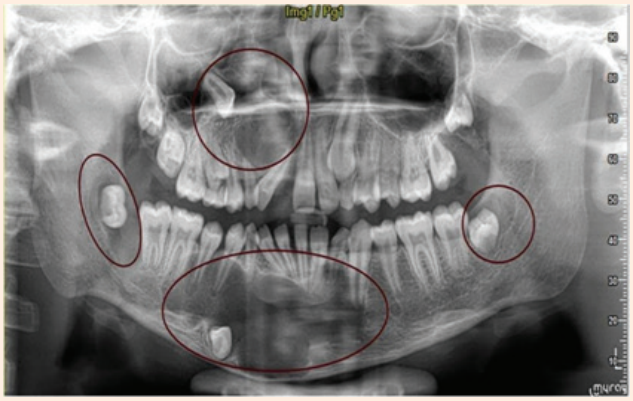

Figure 2: OPG showing multiple radiolucencies.

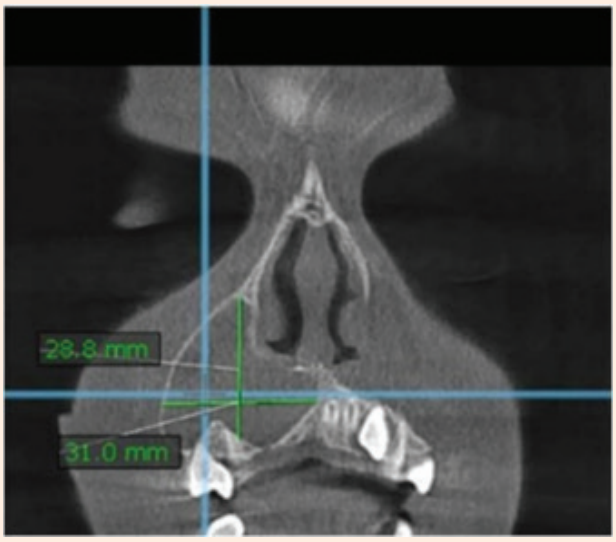

Figure 3: CBCT of maxilla showed a well-defined radiolucency with buccal cortical plate thinning and expansion, displacing the floor of maxillary sinus with impacted 13.

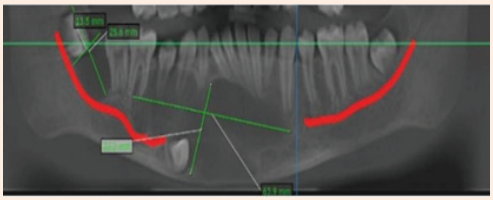

Figure 4: Multiple radiolucencies in the mandible.

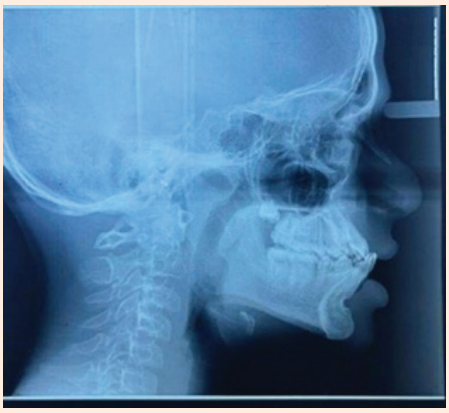

Figure 5: Frontal bossing.

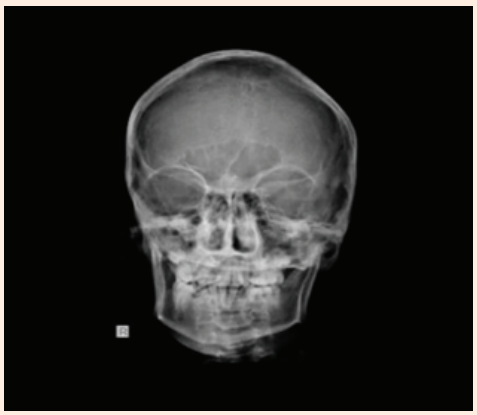

Figure 6: Calcification of flax cerebri.

Incisional biopsy was performed under local anaesthesia, yielding white cheesy material. Histopathological examination confirmed a parakeratinized odontogenic keratocyst (Figure 7).

Figure 7: Aspirated cheesy white material.

Under general anaesthesia and following standard aseptic protocols, including povidone iodine preparation and sterile draping, local infiltration with 2% lignocaine hydrochloride containing adrenaline (1:80,000) was administered in both maxillary and mandibular regions to achieve haemostasis and supplemental analgesia. A crevicular incision was placed in the maxilla extending from the midline to the region of tooth 14, and a mucoperiosteal flap was carefully elevated to expose the lesion. Enucleation and curettage of the cystic cavity were performed with complete removal of the cystic lining, and the impacted maxillary canine (tooth 13), displaced within the lesion, was extracted. The surgical site was irrigated thoroughly with sterile saline to remove debris and minimize recurrence risk.

In the mandible, an incision was placed approximately 5 mm below the crevicular margin in the anterior region, and a bony window was created to access the cystic cavity. The lesion was enucleated and curetted meticulously, followed by extraction of the impacted mandibular canine (tooth 43). Bilateral mucoperiosteal envelope flaps were then raised in the posterior mandible to access lesions associated with the developing third molars. Enucleation and curettage were performed in relation to teeth 38 and 48, and both impacted teeth were removed. Care was taken to preserve vital structures, particularly the inferior alveolar canal, which was noted to be displaced by the lesion.

To reduce the risk of recurrence, adjunctive therapy with 5 fluorouracil (5 FU) was employed [5]. Gelatin sponges impregnated with 5 FU were placed within all bony cavities following enucleation (Figure 8), targeting residual epithelial cells and thereby lowering the likelihood of recurrence. The surgical sites were closed with resorbable vicryl sutures, ensuring tension free closure. The patient tolerated the procedure well, with uneventful healing observed in the immediate postoperative period. Follow up radiographs demonstrated satisfactory bone fill and remodeling, and at six months and twenty months postoperatively, no evidence of recurrence was noted, confirming the effectiveness of the combined surgical and adjunctive approach (Figures 9 & 10).

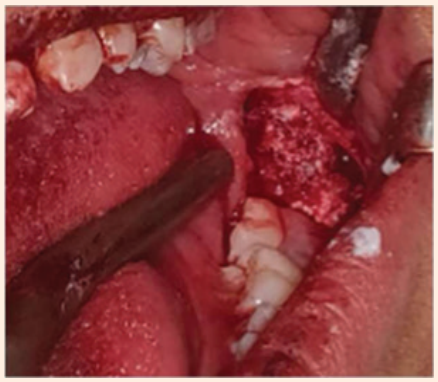

Figure 8: Intraoperative picture.

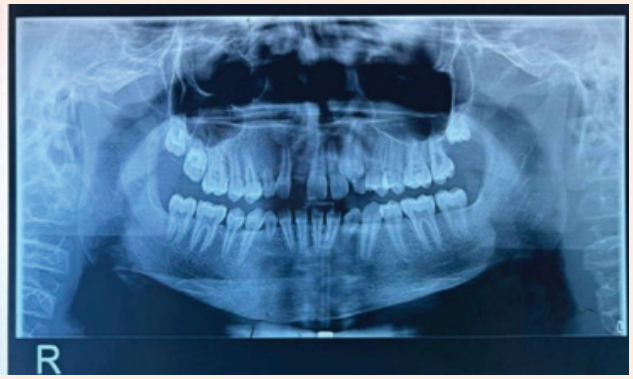

Figure 9: 6 months follow-up OPG.

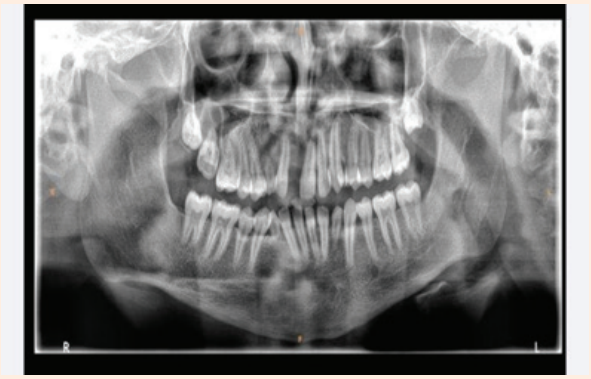

Figure 10: 20 months follow-up OPG.

Discussion

Gorlin–Goltz syndrome, also known as nevoid basal cell carcinoma syndrome (NBCCS), is a rare hereditary condition transmitted in an autosomal dominant pattern with variable expressivity [6]. The syndrome was first described by Gorlin and Goltz in 1960 and has since been recognized as a multisystem disorder with significant oral, skeletal, cutaneous, and systemic manifestations. Its variable penetrance often results in delayed or missed diagnosis, particularly when odontogenic keratocysts (OKCs) are the initial and sole presenting feature.

Odontogenic keratocysts, previously termed keratocystic odontogenic tumors, are frequently the first clinical manifestation of NBCCS [7]. They typically develop during the first, second, and third decades of life [8]. Compared to non-syndromic OKCs, syndromic lesions demonstrate a higher recurrence rate [4]. This increased recurrence has been attributed to several clinicopathological factors, including their large size, cortical perforation, tooth involvement within the cyst lumen, and the presence of daughter cysts [9]. These features highlight the aggressive biological behavior of OKCs in NBCCS and underscore the importance of early recognition. Identification of multiple OKCs in a young patient should immediately raise suspicion for NBCCS, prompting further clinical and radiological evaluation for associated anomalies.

Craniofacial and skeletal anomalies are prominent features of Gorlin–Goltz syndrome. Common findings include bifid, splayed, fused, or misshapen ribs; widened ends of the clavicles; vertebral fusion or hemivertebrae; scoliosis; flame-shaped lucencies of the metacarpals and phalanges; spina bifida occulta; shortened fourth metacarpals; and calcification of the falx cerebri, the latter being recognized as a major diagnostic criterion (10). These skeletal features, although sometimes asymptomatic, provide important diagnostic clues and should be carefully assessed in suspected cases.

Cutaneous manifestations are equally significant in NBCCS. Basal cell carcinomas (BCCs) may appear at a very early age, sometimes as early as two years, and are recognized as one of the major diagnostic criteria [8]. Notably, BCCs in NBCCS can occur in non–sun exposed areas of the body, distinguishing them from sporadic cases. Another cutaneous feature, palmar and plantar pits, is also considered a major diagnostic criterion [12]. These dermatological findings, when present alongside OKCs, strongly support the diagnosis of NBCCS.

In addition to oral, skeletal, and cutaneous features, NBCCS is associated with a wide spectrum of systemic anomalies. These include ovarian fibromas, cardiac fibromas, ophthalmic abnormalities, and medulloblastomas [13]. Such associations necessitate a multidisciplinary approach to patient management, involving oral and maxillofacial surgeons, dermatologists, neurologists, cardiologists, gynaecologists, and genetic counsellors. Early diagnosis and coordinated care are essential to reduce morbidity and improve long-term outcomes.

Management of Gorlin-Goltz syndrome remains challenging. The primary focus is on early detection, conservative surgical management of OKCs, and long-term follow-up. Treatment modalities for OKCs range from marsupialization to enucleation, often supplemented with adjunctive therapies such as peripheral ostectomy, chemical cauterization [14], and topical application of 5-fluorouracil (5 FU) [15]. Carnoy's solution has historically been used as a chemical cauterizing agent, but concerns regarding neurotoxicity have limited its widespread use. Peripheral ostectomy, while effective, may increase morbidity due to the removal of additional bone. In contrast, 5 FU has emerged as a promising adjunctive therapy, selectively targeting residual epithelial cells and reducing recurrence rates with minimal morbidity. Recent studies have demonstrated its efficacy in preventing recurrence after enucleation of OKCs, particularly in syndromic cases.

Given the high recurrence potential of OKCs in NBCCS, long term radiographic surveillance is strongly recommended. Regular follow-up with panoramic radiographs or cone beams computed tomography (CBCT) allows early detection of new or recurrent lesions. Genetic counselling should also be offered to affected families, as NBCCS demonstrates autosomal dominant inheritance with variable penetrance.

Conclusion

Gorlin–Goltz syndrome represents a rare but clinically significant hereditary disorder with wide ranging implications for oral and maxillofacial surgeons. Odontogenic keratocysts often provide the earliest diagnostic clue, particularly in paediatric patients, and their recognition is vital for timely intervention. Surgical management remains the cornerstone of treatment, but the high recurrence potential necessitates adjunctive measures and long term surveillance. Beyond operative care, the surgeon's role extends to identifying syndromic patterns, initiating multidisciplinary collaboration, and guiding patients toward genetic counselling and systemic evaluation. Heightened awareness of the syndrome's diverse manifestations enables clinicians to reduce morbidity, improve quality of life, and optimize long term outcomes. Ultimately, comprehensive evaluation, vigilant follow up, and coordinated interdisciplinary care are essential in managing patients with Gorlin–Goltz syndrome.

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