Case Report

A Vigilant Diagnostic Approach to Gorlin Goltz Syndrome: Case Report

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Abstract

Background: Gorlin-Goltz Syndrome (GGS), or Nevoid Basal Cell Carcinoma Syndrome, is a rare autosomal dominant disorder characterized by odontogenic keratocysts, skeletal abnormalities, and intracranial calcifications. Early diagnosis is crucial for preventing long-term complications and malignancies. **Material and Methods:** A 42-year-old female presented with progressive left maxillary swelling and purulent oral discharge. Clinical examination, radiological investigations, and histopathological analysis were performed to establish the diagnosis. **Results:** Imaging revealed multiple odontogenic keratocysts involving the maxilla and mandible, calcification of the falx cerebri, and bifid ribs. Histopathological examination of the excised maxillary cyst confirmed an odontogenic keratocyst. The presence of three major diagnostic criteria established the diagnosis of Gorlin-Goltz Syndrome. **Conclusion:** This case underscores the importance of careful clinical and radiological evaluation of patients with odontogenic keratocysts. Early recognition of syndromic associations facilitates timely multidisciplinary management and long-term surveillance, thereby reducing disease-related morbidity.

Keywords: Gorlin-Goltz syndrome; Nevoid basal cell carcinoma syndrome; Odontogenic keratocyst; Falx cerebri calcification; Bifid ribs.

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Introduction

Gorlin Goltz Syndrome or Nevoid Basal Cell Carcinoma Syndrome (NBCCS) is a rare genetic disease which is inherited as an autosomal dominant. Gorlin-Goltz Syndrome was first described in 1960 by Robert J. Gorlin and Robert W. Goltz, and is primarily associated with mutations in the PTCH1 gene which is part of the Hedgehog signaling pathway. Gorlin-Goltz Syndrome is characterised by anything from multiple basal cell carcinomas to odontogenic keratocysts, medulloblastomas, palmar and plantar pits, and even skeletal abnormalities, among other things.[1,2]

Early and accurate diagnosis is very important in the management of Gorlin-Goltz Syndrome and in the detection of relatives and care must be taken to ensure that this happens; thorough clinical examination, detailed family history and interpretation of clinical, radiological and genetic testing modalities. The goal of this case report is to emphasize the need for a complete and careful diagnostic work-up of Gorlin-Goltz Syndrome and to outline some of the difficulties and subtleties of its diagnosis and management.[3]

This report aims to enrich the existing published literature on Gorlin-Goltz Syndrome by presenting a detailed case study which outlines the clinical presentation, diagnostic strategies and multidisciplinary management approaches that are required to provide the best care for the patient. The importance of clinicians identifying the subtle signs and symptoms of this syndrome and taking timely action to reduce the long-term effects of the syndrome is highlighted.

Case Presentation

Anamnesis and Physical Examination

A 42-year-old woman was brought to the ENT department complaining of progressive swelling of the left upper jaw for the last two years and continuous discharge from the left upper gum for the past two months. However, she had a previous dental procedure two months before which involved cutting her left sublabial area and she has been seeing excess thick pus discharge since then.

A clinical examination revealed a swelling that was about 3 cm × 2 cm in the left upper region of the jaw [Figure 1]. Swelling was soft to firm, non-tender and non-mobile. Intraoral examination showed a discharging sinus in the left upper gingivolabial sulcus just above the left upper canine, consistent with a chronic inflammatory or a cystic lesion.

An orthopantomogram (OPG) was taken for radiographic assessment and showed bilateral multiple unilocular well-defined radiolucencies in both the mandibular and the maxillary areas

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[Figure 2]. In light of these results, a provisional diagnosis of an odontogenic cyst was made and a further diagnostic evaluation was carried out by using more sophisticated imaging techniques. A detailed contrast enhanced computed tomography (CECT) scan of the face and paranasal sinuses was done with 1-1.5 mm thickness of slices for detailed visualization.

The maxilla and mandible were involved in multiple cystic expansile lesions, which were observed on the CECT scan. In the left maxillary area, there was a large cyst which would strongly suggest a odontogenic keratocyst. An incidental but interesting finding of calcification of the falx cerebri also noted [Figure 3] and have suspected and underlying syndromic association namely Gorlin-Goltz Syndrome. Further to assist with the diagnosis a chest X-ray was performed which confirmed the classic radiologic features of bifid, fused ribs and strengthened the clinical diagnosis of Gorlin-Goltz Syndrome [Figure 4].

Further examination did not show any skin basal cell-like lesions when the skin was examined. Moreover, there was no family history of facial swellings or basal cell nevi.

The patient was operated on for the left maxillary cyst that was causing him symptoms. Cyst enucleation was done through a sublabial approach [Figure 5]. The pathological examination of the removed tissue confirmed the diagnosis and showed the presence of parakeratinized stratified squamous epithelium with palisaded basal cell layer. [Figure 6].

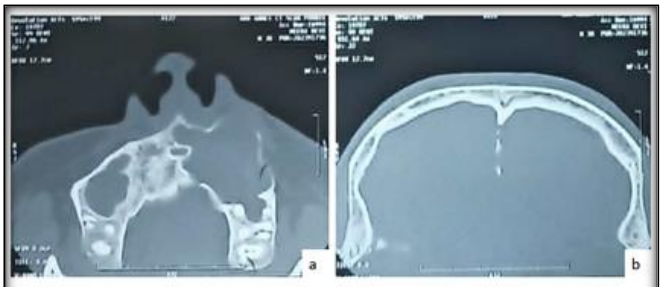

Fig. 3: Axial Computed Tomography a-showing large uniloculated radioluscent lesions in the maxilla; b-showing calcification of falx cerebri.

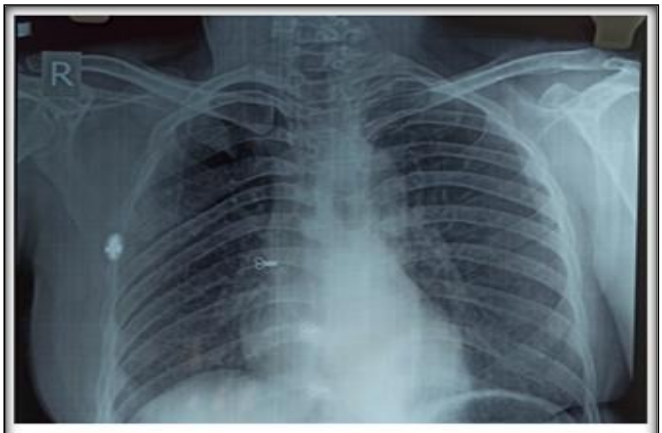

Fig 4. X ray chest showing fused ribs on the right.


Fig 1. Left maxillary swelling

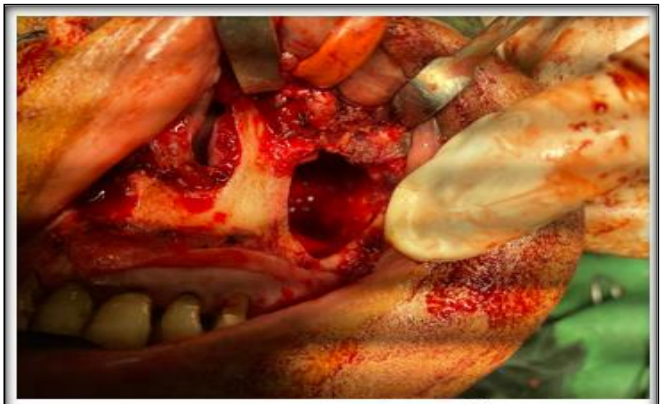
Fig. 5: Intra operative picture- Enucleation of cyst

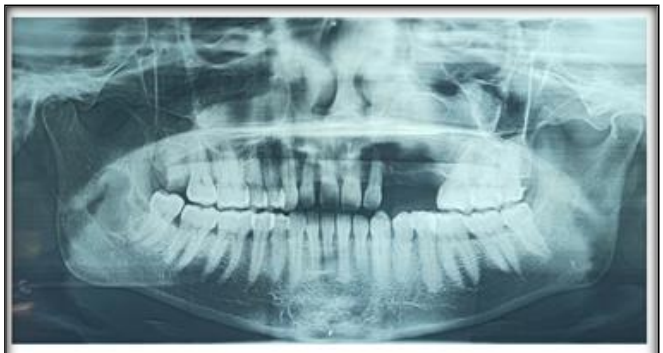
Fig 2. Orthopantomogram showing two maxillary cysts and one cyst in the right hemimandible with absent teeth in left maxilla.

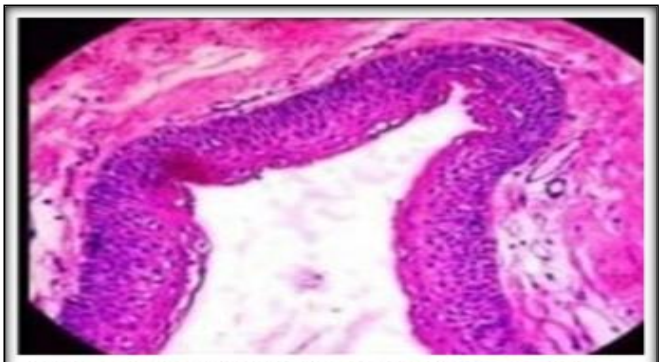
Fig.6: Histopathology examination showing features of an odontogenic keratocyst (H&E, 10X) (Captured on One plus nord)

Discussion

The syndrome of Gorlin-Goltz is a rare, and quite variable, genetic disease. The prevalence of the disease is estimated to be 1 in 57,000 to 1 in 256,000 people.[4] In some cases, an early clinical clue in the diagnosis of odontogenic keratocyst can be the presence of the cysts in the jaws, which tend to occur during the second and third decades of life.[5]

The diagnosis of Gorlin-Goltz Syndrome is made when major and/or minor clinical criteria as defined by Evans et al,[6] (and modified by Kimonis et al,[7] are present. Multiple basal cell carcinomas, odontogenic keratocysts, bifid or fused ribs, intracranial calcifications, and a first degree family member with the syndrome are the major criteria. The minor criteria consist of macrocephaly, cleft lip/palate, skeletal anomalies, ovarian or cardiac fibromas, medulloblastomas. Two major or one major and two minor criteria will confirm a definitive diagnosis. The three major criteria to confirm Gorlin-Goltz Syndrome in the present case were odontogenic keratocysts, bifid ribs, and falx cerebri calcification.

Odontogenic keratocysts associated with Gorlin-Goltz Syndrome demonstrate a higher predilection for the mandible compared to the maxilla, with approximately 69% occurring in the mandibular region and 31% in the maxillary region. Multiple cystic lesions were seen in both the mandible and maxilla, which correspond to the distribution pattern of such lesions as found in affected persons.[8,9]

In Gorlin-Goltz Syndrome, skeletal anomalies are commonly found. Early and common occurrence is the presence of falx cerebri calcification (ca. 85% of the cases). Also, the involvement of the sella turcica is present in 60-80% of cases versus 5% and 4% in the general population. Almost 60% of the cases have rib abnormalities, especially bifid ribs, and 40% have vertebral abnormalities. A short fourth metacarpal (15-40%) is less reliable since the percentage of such digits in the general population is 10%, however, this finding is considered less reliable due to its relatively high occurrence (10%) in the general population.

No cure exists and the goal of management is to detect and prevent complications of Gorlin-Goltz Syndrome early. Basal cell carcinoma needs to be diagnosed and treated at an early stage, so watch for regular dermatologic evaluations. Regular follow-up radiographs or CT scan is very important, and may need to be surgically removed to avoid recurrence. Bifid ribs and kyphoscoliosis are often treated conservatively unless there is severe function impairment. Individuals and families affected are highly recommended to receive genetic counseling services to talk about the risk of inheritance and preventative options.

There is significant morbidity but if early diagnosis and proper management are given, life expectancy is little effected. Targeted molecular therapies including Hedgehog pathway inhibitors (e.g., vismodegib) have been studied as potential approaches to future therapy.[11] The vismodegib (Hedgehog pathway inhibitor) has been studied in the systemic treatment of basal cell carcinoma in GGS patients in a systematic review.[12] In addition, Zhang et al revealed novel mutations in PTCH1 in a Chinese population, providing new insights into the genetic diversity of the disorder.[13]

Eradicating the risks of the syndrome is the most important step to take. These include the possibility of development of neoplasms as a consequence of sun exposure or radiation exposure and, recurrent odontogenic keratocysts, which require long-term monitoring and comprehensive treatment.

The present case highlights the importance of timely diagnosis and management of the Gorlin-Goltz Syndrome by considering multidisciplinary approach and the involvement of otolaryngologists, oral and maxillofacial surgeons, dermatologists, radiologists, and geneticists. The syndrome has a large morbidity associated with its manifestation which can be decreased, and patient's outcomes improved by increased awareness and early identification of the syndrome.

Conclusion

The case report emphasizes the need for a thorough diagnostic work-up of patients who present with odontogenic keratocysts. Incidental calcification of falx cerebri and bifid ribs was noted and the diagnosis of Gorlin-Goltz Syndrome was suggested, highlighting the importance of imaging and careful clinical observation for the diagnosis of syndromic disease. Early diagnosis will help to ensure timely action for follow-up and management thereby avoiding escalation of complications. Treatment of patients affected by this complicated disorder requires a team approach between multiple disciplines.

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Conflicts of interest

There are no conflicts of interest.

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