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GORLIN-GOLTZ SYNDROME THROUGH THE LENS OF IMAGING: A CASE-BASED INSIGHT

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ABSTRACT

Gorlin-Goltz syndrome (GGS), also known as nevoid basal cell carcinoma syndrome (NBCCS), is an autosomal dominant disorder caused by mutations in the PTCH1 gene, located on chromosome 9q22.3. The syndrome is characterized by a triad of multiple basal cell carcinomas (BCCs), odontogenic keratocysts (OKCs), and skeletal anomalies. The incidence of GGS is estimated at 1 in 50,000 to 150,000 individuals, with equal prevalence in males and females [1].
Radiological imaging, including computed tomography (CT) and magnetic resonance imaging (MRI), plays a pivotal role in diagnosing GGS by identifying characteristic features such as calcifications of the falx cerebri, bifid ribs, and OKCs. Early diagnosis is critical to mitigate complications such as malignant transformation of BCCs and maxillofacial deformities secondary to OKCs. The diagnostic criteria for GGS have been refined through international consensus, as outlined by Bree et al. [1], ensuring accurate identification and management of this complex syndrome.

KEYWORDS

Gorlin-Goltz syndrome, nevoid basal cell carcinoma syndrome, odontogenic keratocyst, basal cell carcinoma, skeletal anomalies, falx cerebri calcification, tentorium cerebelli calcification.

MAIN ARTICLE

Introduction

Gorlin-Goltz syndrome (GGS), also known as nevoid basal cell carcinoma syndrome (NBCCS), is an autosomal dominant disorder caused by mutations in the PTCH1 gene, located on chromosome 9q22.3. The syndrome is characterized by a triad of multiple basal cell carcinomas (BCCs), odontogenic keratocysts (OKCs), and skeletal anomalies. The incidence of GGS is estimated at 1 in 50,000 to 150,000 individuals, with equal prevalence in males and females [1].

Radiological imaging, including computed tomography (CT) and magnetic resonance imaging (MRI), plays a pivotal role in diagnosing GGS by identifying characteristic features such as calcifications of the falx cerebri, bifid ribs, and OKCs. Early diagnosis is critical to mitigate complications such as malignant transformation of BCCs and maxillofacial deformities secondary to OKCs. The diagnostic criteria for GGS have been refined through international consensus, as outlined by Bree et al. [1], ensuring accurate identification and management of this complex syndrome.

Case Presentation:

A 50-year-old male presented to the dermatology department with multiple cutaneous lesions and a history of mandibular swelling. Clinical examination revealed multiple basal cell carcinomas and facial asymmetry.

The patient underwent comprehensive radiological evaluation, including cerebral and cervical computed tomography and thoracic computed tomography. All imaging studies were performed at our institution using a 64-slice computed tomography scanner (Siemens Healthcare, Erlangen, Germany).

Diagnostic criteria for Gorlin-Goltz syndrome, as established by Kimonis et al. [2], were applied to confirm the diagnosis. These criteria require the presence of two major or one major and two minor criteria, with major criteria including odontogenic keratocysts, basal cell carcinomas, palmar/plantar pits, and skeletal anomalies.

Radiological examination revealed several characteristic features of Gorlin-Goltz syndrome. Cerebral and cervical computed tomography demonstrated calcifications of the falx cerebri and tentorium cerebelli, which are pathognomonic for this syndrome (Figure 1). Multiple osteolytic lesions consistent with odontogenic keratocysts were identified in the mandible and maxilla (Figure 2A). The patient exhibited hypertelorism with an interorbital distance of 3.9

cm. Multiple dermal lesions with microcalcifications were observed in the nasolabial, buccal, and cervical regions (Figure 2B).

Thoracic computed tomography identified a bifid third left rib, another major criterion for Gorlin-Goltz syndrome (Figure 3). Additionally, micronodules were observed in the lungs, predominantly in the right lower lobe.

Based on these findings, the patient met four major criteria—odontogenic keratocysts (OKCs), calcifications of the falx cerebri, bifid rib, and basal cell carcinomas (BCCs)—as well as one minor criterion (hypertelorism), confirming Gorlin-Goltz syndrome.

The patient underwent surgical excision of the mandibular cysts under general anesthesia, with histopathological confirmation of odontogenic keratocysts. Larger basal cell carcinomas (>1 cm) were excised with 5-mm margins, while smaller lesions (<1 cm) were treated with topical 5-fluorouracil cream (5% twice daily for 6 weeks). Postoperative recovery was uneventful, with significant improvement in facial asymmetry and no signs of cyst recurrence at the 6-month follow-up. Regular surveillance included biannual dermatological exams and annual maxillofacial CT scans to monitor for new lesions.

The management plan included surgical excision of the mandibular cysts, dermatological management of basal cell carcinomas, and regular follow-up for early detection of new lesions and malignancies.

Discussion

Gorlin-Goltz syndrome is a multisystemic disorder with diverse clinical and radiological manifestations. The radiological features of this syndrome are particularly important for early diagnosis and management.

Cerebral imaging typically reveals calcifications of the falx cerebri and tentorium cerebelli, which are considered pathognomonic for Gorlin-Goltz syndrome [3]. These calcifications often develop during the second decade of life and are present in approximately 65–90% (en-dash for numerical range) of patients [4].

Maxillofacial imaging reveals OKCs, which appear as osteolytic lesions in the mandible and maxilla. These cysts typically present as unilocular or multilocular radiolucencies with well-defined sclerotic margins. OKCs in Gorlin-Goltz syndrome have a high recurrence rate of approximately 60% (numerical for percentage), necessitating close radiological surveillance [5].

Thoracic imaging often demonstrates skeletal anomalies, particularly bifid ribs, which are present in about 30–60% (en-dash) of patients with Gorlin-Goltz syndrome [4]. Other skeletal anomalies include scoliosis, vertebral fusion, and shortened metacarpals.

The management of Gorlin-Goltz syndrome requires a multidisciplinary approach. Surgical management of OKCs involves enucleation with or without adjuvant therapies to reduce recurrence. Dermatological management includes regular skin examinations and early treatment of BCCs. Radiological surveillance is essential to monitor disease progression and detect new lesions.

The limitations of this case report include its single-patient focus and the lack of long-term follow-up data. Future research should focus on the correlation between radiological findings and genetic mutations in Gorlin-Goltz syndrome.

Conclusion

Gorlin-Goltz syndrome is a rare but clinically significant disorder with diverse radiological manifestations. Early diagnosis through imaging is crucial for effective management and prevention of complications. Radiologists play a pivotal role in recognizing the characteristic features of Gorlin-Goltz syndrome, particularly in cases presenting with maxillofacial and skeletal anomalies.

Regular radiological surveillance is essential for patients with Gorlin-Goltz syndrome to monitor disease progression and detect new lesions. A multidisciplinary approach involving radiologists, dermatologists, oral surgeons, and geneticists is recommended for optimal management of this complex syndrome.

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Ethics approval and consent to participate

Ethical approval was waived by the institutional review board as this retrospective case report did not involve interventional research.

Consent for publication

Written informed consent was obtained from the patient's legal guardian for publication of this case report and accompanying images. A copy of the consent form is available for review by the Editor-in-Chief.

Availability of data and materials

No datasets were generated or analyzed for this study. Data sharing is not applicable.

Competing interests

The authors declare no competing interests.

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Authors' contributions

- A.B. and L.B.: Manuscript drafting, case analysis, and imaging interpretation.
- S.E.H.: Critical revision and intellectual input.
- N.A. and L.C.: Study supervision and final approval.

All authors read and approved the final manuscript and attest to the accuracy of the data.

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