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A Diagnostic Puzzle in the Jaws: Multifocal Recurrent Odontogenic Keratocysts-Gorlin-Goltz Syndrome

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Abstract: ***Gorlin–Goltz syndrome (GGS) is a rare autosomal-dominant disorder characterized by multiple odontogenic keratocysts (OKCs) and various cutaneous and skeletal abnormalities. A 25-year-old man presented with recurrent posterior mandibular pain and a history of three previous surgeries for jaw lesions. Imaging revealed multifocal maxillary and mandibular radiolucencies, with histopathology confirming OKCs. Palmar/plantar pits and a bifid rib supported the diagnosis despite the absence of basal cell carcinomas, falx calcification, and family history. This case highlights the importance of evaluating recurrent or multifocal OKCs for underlying GGS and ensuring long-term surveillance.***

Keywords: Odontogenic Cysts; Basal Cell Nevus Syndrome; Mandibular radiolucencies; Maxillary radiolucencies; Recurrences

1. Introduction

Odontogenic keratocyst (OKC) is a benign but locally aggressive cystic lesion of the jaws arising from dental lamina remnants, with a well-documented propensity for cortical destruction and recurrence.[1] Although most OKCs are solitary and sporadic, multifocal or bilateral OKCs are a recognised signs for nevoid basal cell carcinoma syndrome (NBCCS), or Gorlin–Goltz syndrome, an autosomal dominant disorder of the PTCH1/SUFU signalling pathway.[2,3] Because jaw cysts frequently precede cutaneous basal cell carcinomas by a decade or more, the dentist or oral and maxillofacial surgeon is often the first clinician positioned to detect NBCCS.[3] We report a young man with thrice-recurrent, synchronous bilateral maxillomandibular OKCs and palmoplantar pitting who underwent repeated surgery over three years without syndromic evaluation, illustrating a diagnostic pitfall and reinforcing the role of the Evans criteria in screening.

2. Case Report

A 25-year-old male presented with mild, dull aching pain in the right posterior mandible and a history of three previous surgeries for similar cystic lesions, with the first diagnosed at 22 years. Clinical examination revealed multiple palmar and plantar pits (Fig.2), while OPG (Fig.2) and CBCT demonstrated extensive multifocal radiolucent lesions involving the maxilla and mandible with associated tooth displacement and cortical expansion. Histopathological examination confirmed odontogenic keratocyst (OKC). Skeletal assessment revealed a bifid rib (Fig.3), and the combination of recurrent multifocal OKCs, palmoplantar pits and bifid rib supported a diagnosis of Gorlin–Goltz syndrome. There was no family history, basal cell carcinoma, falx cerebri calcification, macrocephaly, cleft lip/palate, or ovarian/cardiac fibroma. The patient underwent surgical management under general anaesthesia. The left maxillary OKC was treated by enucleation through a lateral bony-window approach, with removal of the associated impacted teeth (Fig.5), repositioning and fixation of the bony segment using a 1.5-mm plate and screws, and primary closure. The right mandibular lesion was managed by extraction of 47, enucleation with bone guttering, and Carnoy's solution followed by saline irrigation. Postoperative recovery was uneventful, and long-term clinical and radiographic surveillance was advised due to the recurrent nature of syndromic OKCs.

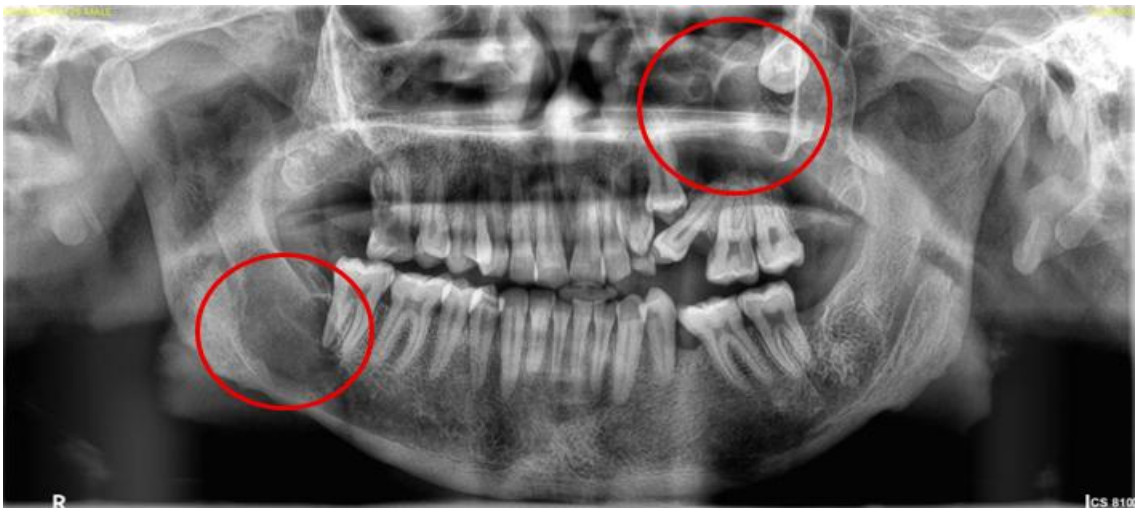

Figure 1

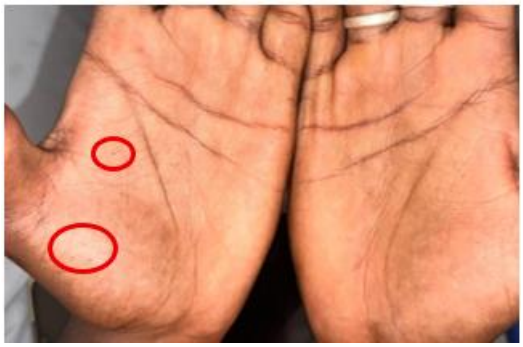

Figure 2

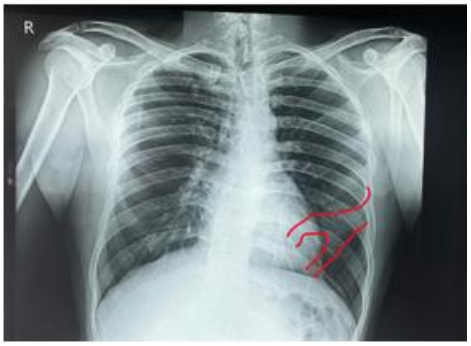

Figure 3

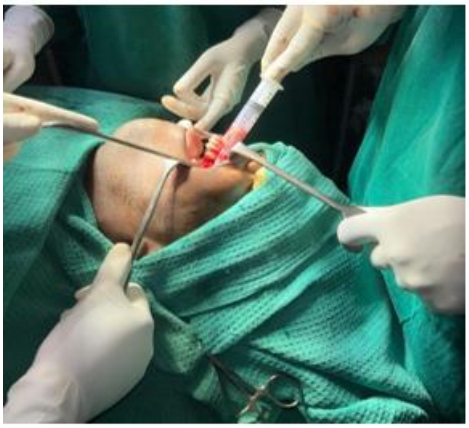

Figure 4

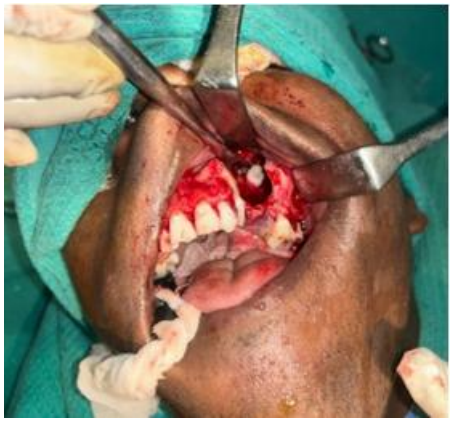

Figure 5

3. Discussion

OKCs account for approximately 10% of jaw cysts and, following the WHO's return to cystic rather than neoplastic terminology, are again labelled odontogenic keratocysts rather than keratocystic odontogenic tumour.[4] They occur most frequently in the posterior mandible and are often discovered incidentally. These lesions commonly exhibit infiltrative buccolingual expansion, with recurrence rates of 17–56% following simple enucleation. However, recurrence rates can fall below 2% when enucleation is combined with adjunctive Carnoy's solution or peripheral ostectomy, as supported by recent network meta-analyses and scoping reviews.[5]

Multiple or recurrent OKCs occurring in the first three decades of life should always raise suspicion for NBCCS. Evans et al. proposed diagnostic criteria requiring two major, or one major and two minor, features; major criteria include multiple basal cell carcinomas, histologically proven OKC, three or more palmar/plantar pits, lamellar falx calcification, bifid/fused ribs, and a first-degree relative with NBCCS.[2] Our patient met two major criteria – multifocal, recurrent OKC and palmoplantar pitting- sufficient for a syndromic diagnosis even before cutaneous basal cell carcinomas, falx calcification, or a family history had emerged, consistent with the well-recognised observation that jaw cysts are usually the earliest sign of NBCCS.[3]

This case highlights a genuine diagnostic pitfall: three prior surgeries over three years were performed for 'recurrent cysts' without the palmoplantar pits – a simple, cost-free bedside sign – ever prompting syndromic evaluation. Systematic screening at first presentation of a multifocal or recurrent jaw cyst, using the Evans or the subsequently refined Kimonis criteria, would have allowed earlier diagnosis, targeted counselling regarding lifelong basal cell carcinoma and, in PTCH1/SUFU carriers, medulloblastoma risk, and closer radiographic surveillance.[3] Confirmed NBCCS mandates a multidisciplinary team – oral and maxillofacial surgery, dermatology, ophthalmology, clinical genetics and radiology – together with genetic counselling for first-degree relatives given autosomal dominant inheritance with high penetrance.[23]

Management of the jaw lesions did not differ from sporadic OKC: enucleation with adjunctive Carnoy's solution or peripheral ostectomy remains the favoured balance of recurrence control and morbidity, with resection reserved for extensive or repeatedly recurrent disease.[5] However, because NBCCS-associated OKCs recur more often and at multiple sites, surveillance should be closer and lifelong, with a low threshold for repeat imaging of new asymptomatic lesions. A limitation of this report is that formal histopathological confirmation and genetic testing were pending at the time of writing; nonetheless, the clinical and radiological picture, combined with the pathognomonic cutaneous finding, was considered sufficient to flag syndromic disease and initiate work-up without delaying necessary surgery.

Tables and Legends

Table 1: Evans criteria for nevoid basal cell carcinoma syndrome applied to the present patient

Criterion type	Feature	Present in this patient
Major	Multiple basal cell carcinomas (or 1 if age <20 y)	Not present
Major	Odontogenic keratocyst of the jaw	Present
Major	≥3 palmar or plantar pits	Present
Major	Lamellar calcification of falx cerebri	Not assessed – skull imaging planned
Major	Bifid rib	Present
Major	First-degree relative with NBCCS	Not elicited
Minor	Macrocephaly	Absent
Minor	Cleft lip/palate, frontal bossing, hypertelorism	Absent
Minor	Sprengel deformity, pectus deformity, syndactyly	Absent
Minor	Bridging of sella turcica / vertebral anomalies	Not assessed
Minor	Ovarian fibroma	Not applicable (male)
Minor	Medulloblastoma	Absent

Diagnosis of NBCCS requires two major, or one major plus two minor, criteria (Evans et al.[2]). This patient fulfilled two major criteria – multifocal/recurrent odontogenic keratocyst and ≥3 palmoplantar pits – supporting a working diagnosis pending completion of falx-calcification imaging, family screening and histopathological/genetic confirmation.

4. Conclusion

Synchronous or recurrent multifocal odontogenic keratocysts, especially in young patients, warrant a deliberate search for palmoplantar pits and other Evans/Kimonis criteria before each episode is treated as an isolated surgical problem. Prompt recognition of NBCCS changes counselling, follow-up intensity and family screening, and the dental or maxillofacial surgeon is frequently the first, and sometimes only, clinician positioned to make this diagnosis.

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