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Odontogenic Keratocyst in Syndromic and Recurrent Sporadic Forms: Two Case Reports

Ghada Rekik[1] , Marwa Garma[1] , Chaima Khalifa[1] , Afef Slim[1], Abdellatif Chokri[1], Hajer Hentati[1]

1-Department of oral medicine and oral surgery, Academic dental clinic of Monastir, university of Monastir, Tunisia, laboratory of oral health and maxillofacial rehabilitation (LR12ES11)

Corresponding author:

Ghada Rekik

E-mail : ghadarekik1997@gmail.com

Abstract

Background: Odontogenic keratocyst (OKC) is a developmental odontogenic cyst characterized by locally aggressive behavior and a high recurrence rate. Its clinical and radiological similarity to other odontogenic cysts frequently leads to diagnostic errors. Moreover, its association with nevoid basal cell carcinoma syndrome (Gorlin-Goltz syndrome) adds complexity to diagnosis and management.

Case Reports: Two cases of odontogenic keratocyst highlighting distinct diagnostic challenges. The first case concerns a recurrent OKC initially misdiagnosed as a residual cyst and treated without histopathological confirmation, resulting in delayed diagnosis and recurrence. The second case involves a 22-year-old female referred for multiple jaw radiolucencies, in whom the diagnosis of odontogenic keratocyst led to the identification of Gorlin-Goltz syndrome.

Discussion: These cases emphasize the importance of systematic histopathological examination of all jaw cysts, careful evaluation of recurrent or atypical lesions, and consideration of syndromic associations in young patients or in the presence of multiple lesions. Accurate diagnosis, appropriate treatment, and long-term follow-up remain essential for optimal management of odontogenic keratocysts.

Keywords : Odontogenic keratocyst; Residual cyst; Recurrence; Gorlin-Goltz syndrome; Odontogenic cysts.

Introduction

Odontogenic keratocyst (OKC) is classified as a developmental odontogenic cyst according the world health organisation 2022[1,2], known for its locally aggressive behavior and high recurrence potential. Despite its benign nature, OKC continues to represent a diagnostic and therapeutic challenge in clinical practice.

Clinically and radiographically, OKC may present with nonspecific features and often mimics other odontogenic cysts, such as residual cysts or dentigerous cysts, as well as odontogenic tumors. This overlap may lead to misdiagnosis and inappropriate initial management. Histopathological examination remains the cornerstone of diagnosis, typically revealing a parakeratinized stratified squamous epithelial lining with a palisaded basal cell layer and a thin, friable cyst wall[6,7].

OKC may occur as an isolated lesion or in association with nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin-Goltz syndrome, an autosomal dominant disorder commonly linked to mutations in the *PTCH1* gene. In syndromic cases, OKCs tend to occur at a younger age, are frequently multiple, and show an increased risk of recurrence. Importantly, odontogenic keratocysts may represent the first clinical manifestation of the syndrome, placing dental practitioners in a key position for early diagnosis[8,9].

This motivated the present work, which aimed to report two cases of odontogenic keratocyst illustrating distinct diagnostic challenges : a recurrent lesion initially misdiagnosed as a residual cyst, and a syndromic case in which multiple jaw lesions led to the diagnosis of Gorlin-Goltz syndrome in order to highlight their diagnostic and therapeutic approachs, and to emphasize the role,of dentists in detecting general diseases .

Case Reports

A 50-year-old patient was referred to the oral medecine and oral surgery department at the medical dentistry clinic of Monastir , Tunisia, for evaluation of a recurrent radiolucent lesion in the posterior mandibular region. The patient’s medical history was notable for cardiovascular disease with left ventricular hypertrophy, lymphoedema, and rheumatoid arthritis under long-term immunosuppressive therapy. No facial asymmetry or sensory disturbances were reported at the time of presentation.

The patient had previously undergone dental extractions in the affected area several years earlier, after which a radiolucent lesion was detected and diagnosed as a residual cyst. Surgical enucleation was performed without histopathological confirmation.

Clinical follow-up was irregular, and no long-term surveillance was established.

At the current presentation, Extraoral examination revealed no facial asymmetry, swelling, or cutaneous abnormalities . However, the intraoral examination revealed a vestibular swelling in the posterior mandibular region, exhibiting a characteristic “ping-pong ball” sensation on palpation, suggestive of a cystic lesion with cortical bone thinning.(figure 1a; 1b).

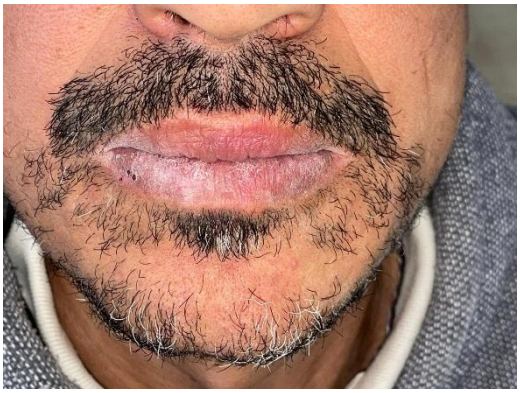

Figure 1.a. Extraoral examination

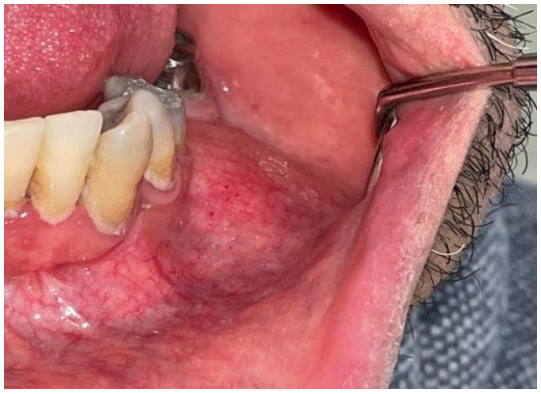

Figure 1.b. Intraoral examination

Radiographic evaluation using panoramic radiography demonstrated a well-defined radiolucent image in the posterior mandible, extending along the previous extraction site (figure 2.a). Cone-beam computed tomography (CBCT) revealed a unilocular osteolytic image with well-corticated margins, showing anteroposterior extension with minimal buccolingual expansion, findings suggestive of an odontogenic lesion (figure 2.b).

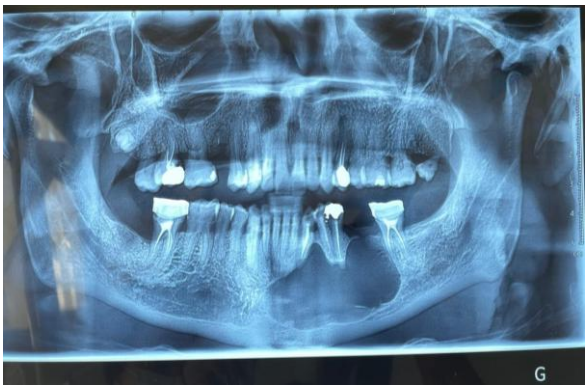

Figure 2.a

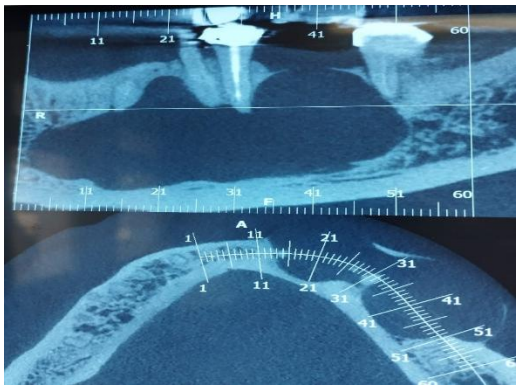

Figure 2.b

Figure2. Radiographic appearance: 2.a Panoramic radiograph showing a well-defined radiolucent image in the posterior mandible, consistent with a cystic pathology. 2.b CBCT images demonstrated a well-circumscribed hypodense lesion in the posterior mandible with areas of cortical thinning and focal cortical perforation.

Given the history of recurrence and radiological features, surgical exploration was undertaken. Complete enucleation of the lesion was performed. During surgical exploration, a thick, whitish, keratin-like material was observed to drain from the lesion, a characteristic finding highly suggestive of odontogenic keratocyst. This intraoperative feature reflects the keratinized epithelial lining of the cyst and supports both the diagnosis and the long-standing nature of the lesion. (figure 4.a; 4.b)

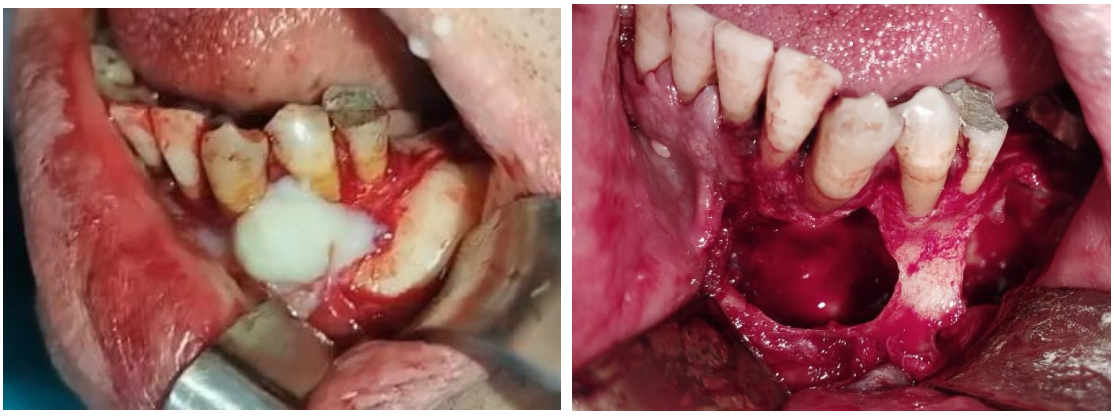

Figure 3.a. **Figure 3.b.**

Figure 3. Intraoperative finding
3. a. Intraoperative view showing drainage of thick, white keratinous material. 3. b. Surgical view showing complete enucleation of the cystic lesion with careful removal of the cystic lining.

The specimen was systematically submitted for histopathological examination. Microscopic analysis revealed a cystic lesion lined by a parakeratinized stratified squamous epithelium with a palisaded basal cell layer and a corrugated surface, consistent with the diagnosis of odontogenic keratocyst.

Based on the definitive diagnostic, the patient was enrolled in a strict postoperative follow-up protocol. Clinical healing was uneventful, with complete resolution of the vestibular swelling and absence of pain or signs of infection. Radiographic follow-up demonstrated progressive bone regeneration at the surgical site, with no evidence of residual or recurrent lesion. (Figure4.a.; 4.b.)

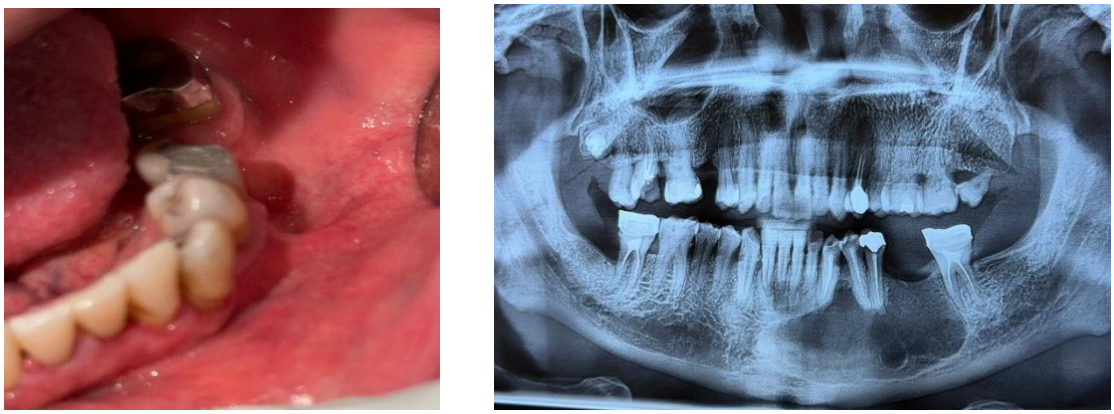

Figure4. a. **Figure4. b.**

Figure 4. Six-month postoperative follow-up. Figure 4.a. Clinical healing was uneventful. Figure 4.b. Panoramic radiograph showing progressive bone regeneration at the surgical site with no evidence of recurrence

The second case is about a 19-year-old female was referred following the incidental discovery of multiple radiolucent images on an orthopantomogram (OPG) performed during routine dental evaluation. The patient was asymptomatic and reported no pain or swelling. Her medical history was unremarkable.

Extraoral examination revealed hypertelorism, with normal facial proportions and no evidence of facial asymmetry or swelling. The overlying skin appeared normal, and no cervical lymphadenopathy was detected.

General physical examination showed multiple pigmented cutaneous lesions resembling nevi, distributed over the face, trunk, and upper extremities, including the palms (figure 5). These lesions were well-defined, variable in size, and consistent with melanocytic nevi, raising the possibility of a syndromic condition such as nevoid basal cell carcinoma syndrome.

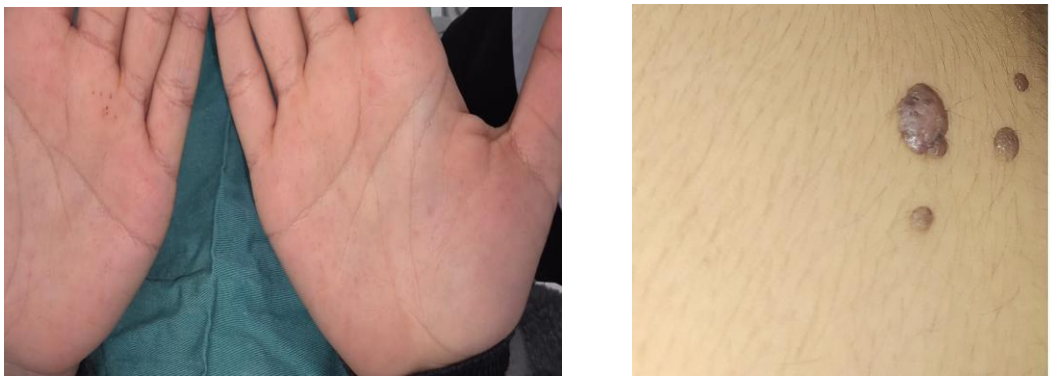

Figure 5. Cutaneous examination

Multiple well-defined pigmented nevi distributed over palms and body skin, suggestive of a syndromic association such as Gorlin-Goltz syndrome.

Intraoral examination demonstrated normal oral mucosa, with no signs of inflammation, ulceration, or mucosal pigmentation. The dentition was within normal limits, with no vestibular obliteration or cortical expansion observed. (figure 7)

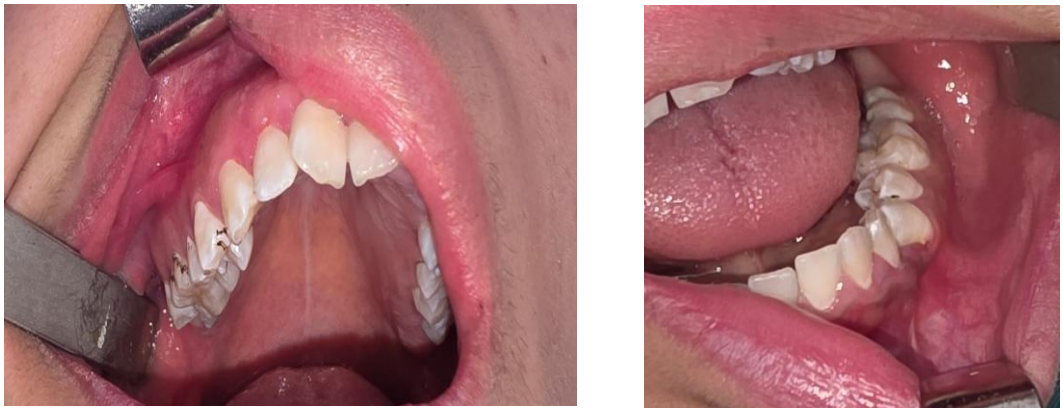

Figure 6. Intraoral examination

Intraoral view showing normal mucosa without swelling or inflammation.

The combination of palmar pigmentation, multiple nevi, and hypertelorism suggests the need for further investigation for a potential syndromic association.

Panoramic radiograph revealed multiple well-defined radiolucent images affecting different regions of the jaws. CBCT confirmed the presence of several osteolytic lesions with well-corticated margins, showing predominantly anteroposterior extension and minimal buccolingual expansion.

Surgical enucleation of one maxillary lesion was performed for diagnostic confirmation. (figure7)

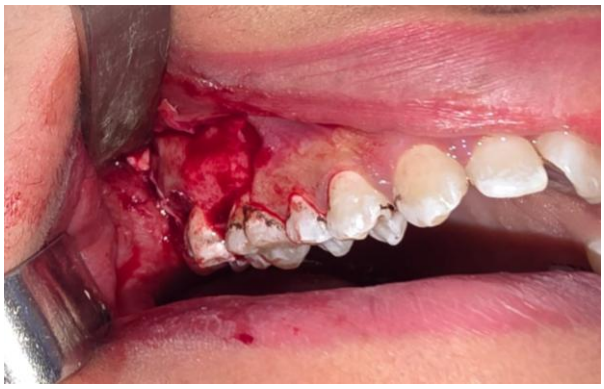

Figure 7. Intraoperative view

Enucleation of the maxillary lesion with the impacted upper right third molar .

Histopathological examination demonstrated features characteristic of odontogenic keratocyst. Considering the patient's young age, multiplicity of jaw lesions, and cutaneous findings, further investigations were undertaken. The clinical findings fulfilled the diagnostic criteria for nevoid basal cell carcinoma syndrome (Gorlin-Goltz syndrome).

The patient was referred for multidisciplinary management, including dermatologic and genetic evaluation. Conservative surgical management of the remaining lesions was planned, with strict long-term clinical and radiological follow-up.

Disussion

Odontogenic keratocyst (OKC) is a developmental odontogenic cyst first described in the 1950s and historically considered a "keratocystic odontogenic tumor" due to its aggressive behavior and high recurrence potential. The World Health Organization (WHO) classification of 2022 now places OKC among developmental odontogenic cysts, emphasizing its local aggressiveness, tendency for recurrence, and its origin from dental lamina remnants [1,2]. OKCs are classified as sporadic (isolated) lesions or as part of a syndromic presentation, most commonly associated with nevoid basal cell carcinoma syndrome (NBCCS or Gorlin-Goltz syndrome) [3,4]. This distinction is critical because syndromic OKCs often present with multiple lesions, occur at younger ages, and may recur more frequently, necessitating broader clinical surveillance.

The clinical presentation of OKCs varies according to their form. Sporadic OKCs typically manifest as solitary lesions, frequently located in the posterior mandible, although maxillary involvement can also occur. Clinically, they are often asymptomatic and may be discovered incidentally during routine radiographs. When larger, they can cause swelling, pain, tooth displacement, or cortical expansion. Syndromic OKCs, by contrast, frequently present as multiple cysts, appearing at an earlier age, and are often accompanied by systemic features such as palmar or plantar pigmentation, hypertelorism, skeletal anomalies, and, in some cases, basal cell carcinomas [3,4]. In our second case, multiple pigmented nevi involving the face, trunk, and palms, together with hypertelorism, raised suspicion of NBCCS. Recognition of these systemic features is essential for early diagnosis and holistic patient management. Radiographically, OKCs usually present as well-defined radiolucent lesions, which can be unilocular or multilocular. Borders may appear smooth or scalloped, and lesions can cause displacement of adjacent teeth or thinning of cortical bone without significant expansion. Root resorption is uncommon but may occur in larger lesions [5]. Histologically, OKCs are characterized by a parakeratinized stratified squamous epithelial lining, typically 6-10 cells thick, with a palisaded basal layer and corrugated surface. The connective tissue wall is generally thin and devoid of inflammation unless secondarily infected. Syndromic OKCs may exhibit more aggressive features, such as satellite cysts or daughter cysts within the wall, contributing to the higher recurrence rate [6,7].
At the molecular level, mutations in the PTCH1 gene, a component of the hedgehog signaling pathway, have been implicated in both sporadic and syndromic OKCs. PTCH1 mutations are more commonly observed in syndromic OKCs and are associated with multiple cyst development and increased recurrence risk. These genetic insights reinforce the importance of careful systemic evaluation in patients presenting with multiple or early-onset OKCs [8,9].
Given the variable presentation of OKCs, accurate diagnosis of OKC requires careful integration of clinical, radiographic, and histopathologic findings. The differential diagnosis of sporadic OKCs includes Dentigerous cysts Often associated with the crown of an unerupted tooth; histologically lined by non-keratinized epithelium, lacking the palisaded basal layer of OKCs. Radicular cysts Typically inflammatory, associated with non-vital teeth, with chronic inflammatory infiltrates in the wall. Ameloblastoma also may mimic multilocular OKCs radiographically but histologically shows epithelial islands with palisaded columnar cells and stellate reticulum-like centers. Other developmental cysts: Such as lateral periodontal cysts or glandular odontogenic cysts, which differ in location, histology, and clinical course [5,6].

In syndromic cases, the presence of multiple lesions, palmar/plantar pigmentation, skeletal anomalies, and facial features such as hypertelorism strongly suggest NBCCS, and systemic screening is recommended. These findings underscore the important role of the dentist not only in managing the cyst itself but also in identifying patients at risk for systemic manifestations, facilitating timely multidisciplinary care [3,4].

The management of OKC requires surgical excision with careful consideration of recurrence risk. Enucleation is commonly employed for smaller lesions, while larger or recurrent cysts may require peripheral ostectomy or adjunctive therapy (e.g., Carnoy’s solution). Follow-up is critical due to the potential for late recurrence, particularly in syndromic patients [7,10].

Dentists play a pivotal role not only in the diagnosis and treatment of the cyst but also in recognizing systemic signs that may indicate an underlying syndrome, enabling timely

referral and multidisciplinary management.

Conclusion

Odontogenic keratocyst is a diagnostically challenging lesion with significant recurrence potential. These two cases underline the importance of accurate diagnosis, systematic histopathological examination, and long-term follow-up. Furthermore, odontogenic keratocysts may represent the first manifestation of Gorlin-Goltz syndrome, emphasizing the critical role of dental practitioners in early detection and multidisciplinary management.

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