

HYBRID AMELOBLASTOMA AND ODONTOGENIC KERATOCYST IN A PATIENT WITH GORLIN-GOLTZ SYNDROME: CASE REPORT

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ABSTRACT

Introduction: Hybrid odontogenic lesions combine characteristics of two or more lesions in a single site, and they are considered a rare condition. The occurrence of these lesions in patients with Gorlin-Goltz syndrome is even less common. Since the lesion has variable clinical aspects and multiple imaging characteristics, histopathological examination is mandatory for a definitive diagnosis. **Methods:** This article reports the case of a patient with Gorlin-Goltz syndrome, who had multiple odontogenic keratocysts in the mandible and maxilla, as well as a combined/hybrid ameloblastoma and odontogenic keratocyst lesion. **Results:** Distinct areas with histological patterns characteristic of the cyst and tumor were observed within the same lesion. **Conclusion:** This unusual but still possible diagnosis should be considered by pathologists and oral and maxillofacial surgeons when evaluating, diagnosing and treating patients with syndromes characterized by odontogenic lesions.

Keywords: *ameloblastoma; odontogenic keratocyst; Gorlin-Goltz Syndrome; hybrid lesion*

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INTRODUCTION

Hybrid odontogenic lesions are defined by the combined histopathological characteristics of cysts and/or odontogenic tumors at the same primary site. The occurrence of hybrid lesions composed of ameloblastoma and odontogenic keratocyst are an extremely rare^{1,2,3,4}.

The odontogenic keratocyst, a locally invasive cystic lesion with a high recurrence rate, is one of the major criteria for the diagnosis of Gorlin-Goltz syndrome (Nevoid basal cell carcinoma syndrome)⁵.

Ameloblastoma is a slow-growing, benign, odontogenic tumor that affect the posterior region of the mandible in most cases. It is an aggressive neoplasm that tends to infiltrate the bone trabeculae⁶.

Usually, both odontogenic keratocyst and ameloblastoma are found on imaging and are difficult to distinguish clinically and radiographically, thus requiring histopathological examination for the final diagnosis. Hybrid odontogenic lesions, which combine histological characteristics of different lesions, present an even greater diagnostic challenge^{1,2,7,8,9}.

The main objective of this article was to describe an unusual case of a combined/hybrid ameloblastoma and odontogenic keratocyst in a patient with Gorlin-Goltz syndrome with multiple keratocysts in the maxilla and mandible.

CASE REPORT

Patient WCV, male, 30 years old, with a genetic diagnosis of Gorlin-Goltz syndrome [karyotype: 46, XY, del (9)(q22.1-22.33)], is currently under clinical care at the Hospital de Clínicas de Porto Alegre (HCPA), presenting with several teeth with coronoradicular destruction due to dental caries. No expansive intra-buccal lesions were observed.

Panoramic radiographic examination (Figure 1) revealed four osteolytic lesions: a radiolucent lesion with undefined limits in the right maxilla,

a well-defined radiolucent lesion in the right posterior mandible, a unilocular, well-defined radiolucent lesion with a radiopaque halo in the left posterior mandible, and a radiolucent lesion with undefined limits in the anterior region of the right mandible. Considering the genetic diagnosis of Gorlin–

Goltz syndrome, the diagnostic hypothesis for lesions in the mandible was multiple odontogenic keratocysts; for maxillary lesions, the hypothesis of an inflammatory apical lesion was considered to be due to the presence of teeth with extensive caries-related lesions.



Figure 1: Panoramic radiography, showing radiolucent lesions (arrows) in the maxilla and mandible. Coronoradicular lesions caused by dental caries are present.

The patient underwent surgical intervention under general anesthesia for the total removal of the intraosseous lesions in the maxilla and mandible, as well as dental extractions. Peripheral osteotomy and liquid nitrogen cryotherapy were performed at all surgical sites as adjuvant therapies. The excised lesions were subjected to histopathological analysis.

The lesion of the right maxilla was characterized by the presence of a cystic cavity coated with a parakeratinized stratified epithelium with a few layers, a corrugated surface, and a basal layer with hyperchromatic nuclei arranged in a palisade. Fragile union between the epithelium and connective tissue, and a flat interface without epithelial crests were observed. Microscopy revealed typical characteristics of odontogenic keratocyst (Figure 2A). In a region distant from the cystic cavity, islands of odontogenic epithelium are separated by dense connective tissue. On some islands, peripheral palisading of cells can be observed; however, in most of them, the cells appear more cuboidal. No stellate reticulum pattern was observed at the center of the islands. The microscopic characteristics resembled those of basal cell ameloblastoma (Figure 2B).

The lesion located in the right posterior mandible was characterized by a cystic cavity covered by a

parakeratinized stratified epithelium consisting of a few layers, with a corrugated surface and a basal layer with hyperchromatic nuclei arranged in a palisade. Fragile union between the epithelium and connective tissue, and a flat interface without epithelial crests were observed. Microscopy revealed odontogenic keratocyst characteristics (Figure 2C).

Microscopic characteristics typical of odontogenic keratocyst were observed in the lesions located in the left posterior mandible (Figure 2D). Some islands of odontogenic epithelium with basal cells and polarized nuclei, such as pre-ameloblasts, which are characteristic of ameloblastoma, were observed in another region of the same lesion. The stellate reticulum pattern at the center of the islands is not well defined (Figure 2E).

The lesion, located in the right anterior mandible, showed a cavity covered with a parakeratinized stratified epithelium and a basal layer with hyperchromatic nuclei. Epithelial cells proliferated towards the connective tissue, forming islands of odontogenic epithelium in a solid pattern (Figure 2F). This feature has been reported in cases of isolated odontogenic keratocyst and in patients with Gorlin–Goltz syndrome.

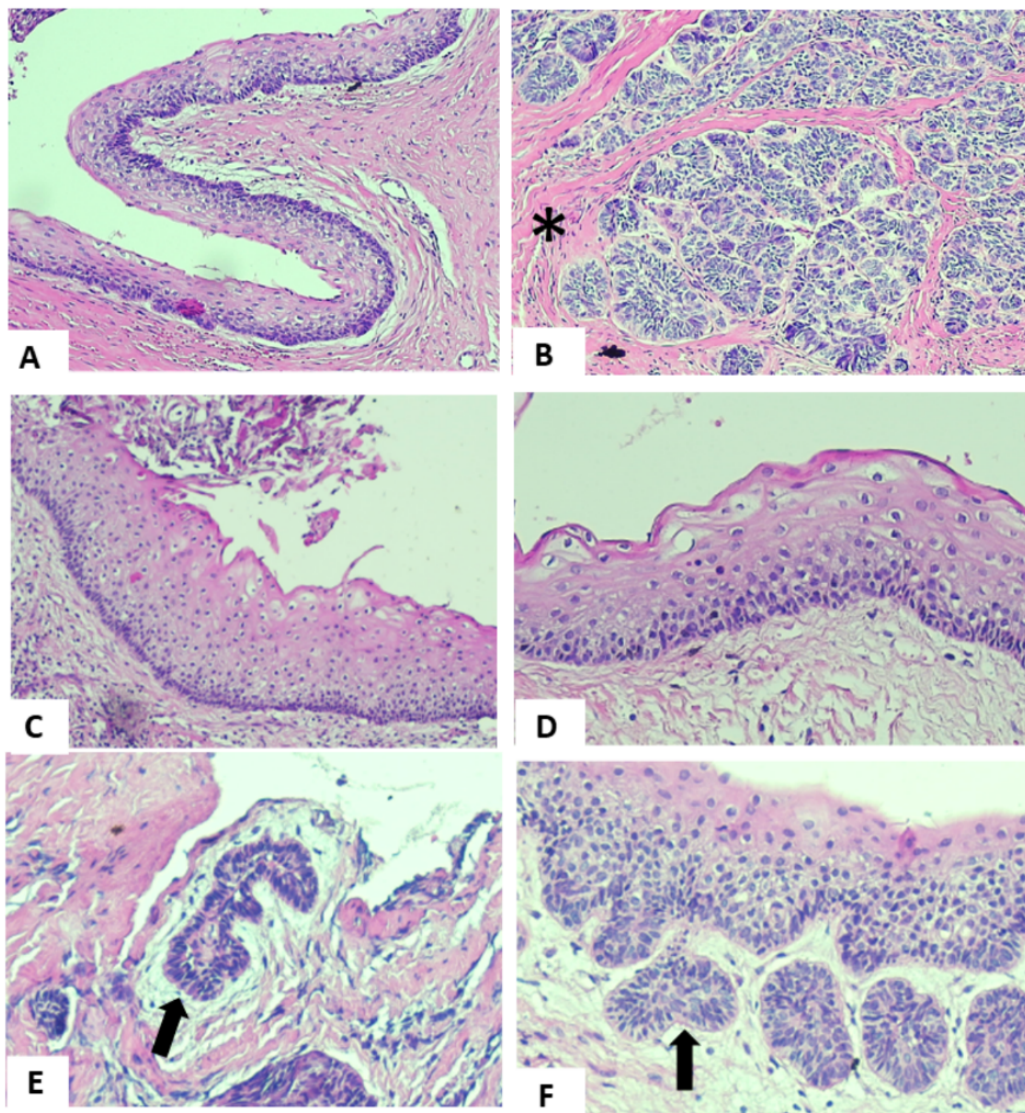


Figure 2: Microscopic aspect of the lesions. A and B: Lesion in the right maxilla; A: microscopic characteristics of keratocysts; B: islands of odontogenic epithelium between fibrous connective tissue (*), characteristic of ameloblastoma. C: Microscopy of the lesion in the right posterior mandible shows an odontogenic keratocyst. D and E: Lesion in the left posterior mandible. D: An odontogenic keratocyst; E: islands of the odontogenic epithelium in the connective tissue capsule, presenting high peripheral cells (arrows) with ameloblastomatous characteristics; F: Lesion in the anterior right mandible, showing the proliferation of islands of odontogenic epithelial cells (arrows) from the lining epithelium of the odontogenic keratocyst.

DISCUSSION

Hybrid odontogenic lesions are extremely rare¹. Hybrid lesions composed of ameloblastoma and odontogenic keratocyst have been described in the literature as keratocyst with ameloblastic alterations consisting of reverse polarity and apical vacuolization^{1,2}. There is an isolated case of simultaneous occurrence of both lesions in the published English literature, but as completely distinct events: odontogenic keratocyst on the left side of the mandible and ameloblastoma on the right side of the mandible⁵. Neuman et al.¹ described two cases

of hybrid odontogenic lesions, one of which was an odontogenic keratocyst presenting as odontogenic epithelial islands with ameloblastic characteristics far from the cystic cavity, as in the present case.

Because the patient was diagnosed with Gorlin–Goltz syndrome, imaging examinations showing osteolytic lesions in the maxilla and mandible were preoperatively interpreted as odontogenic keratocyst⁷. The lesions located in the right posterior and right anterior mandible were histopathologically diagnosed as odontogenic keratocyst with a characteristic histopathological pattern. Histopathological examination

of the lesions excised from the right maxilla and left posterior mandible revealed cystic lesions with characteristics of odontogenic keratocyst and islands of odontogenic epithelium in the stroma. In this case report, the right maxilla presented with two distinct lesions at the same site, which were diagnosed using microscopy as hybrid lesion. The lesion in the left posterior mandible presented as a few islands with an ameloblastic pattern, which may have been an ameloblastomatous alteration in the islands of the odontogenic epithelium from the odontogenic keratocyst. These characteristics have also been described individually or together in other reports on hybrid lesions^{1,7,9,10}.

Odontogenic keratocyst and ameloblastoma may present with similar clinical and imaging features, requiring histopathological examination for a final diagnosis^{3,8,9}. The literature discusses whether hybrid odontogenic lesions originate concomitantly from a common pluripotent epithelial cell, or represent the coexistence of two distinct lesions¹.

The patient was treated with enucleation of the lesions, followed by adjuvant therapy (peripheral ostectomy and cryotherapy). This therapeutic option is recommended for hybrid lesions, because recurrence is rare⁷. The patient was kept under clinical and imaging monitoring, with no signs of lesion recurrence at the five-year postoperative follow-up.

In conclusion, the occurrence of a combined/hybrid odontogenic lesion composed of ameloblastoma and odontogenic keratocyst in a patient with Gorlin–Goltz syndrome with multiple odontogenic keratocystic lesions is rare, and little is about the etiology of the lesion, whether they were originally two separate lesions coexisting or an ameloblastic transformation of an odontogenic keratocyst, as suggested by some microscopic fields (Figure 2F). The diagnosis of hybrid odontogenic lesions should be considered and presents a challenge for oral and maxillofacial surgeons and oral pathologists; because their histogenesis is controversial, imaging results may vary, and clinical behavior is still poorly understood.

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