# Familial Multiple Odontogenic Keratocysts

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## ABSTRACT

Multiple odontogenic keratocysts (OKCs) are principle features of nevoid basal cell carcinoma syndrome (NBCCS; Gorlin-Goltz syndrome). NBCCS is a genetic disorder transmitted by an autosomal dominant gene with variable expressivity, which is important to recognize when a patient has multiple OKCs. The cysts of the jaws are among the most common findings. Another feature is a certain appearance of the face, such as: large calvaria, high-arched eyebrows, broad nasal root, and mild hypertelorism. Before-therapy diagnosis is, therefore, as important as after-therapy diagnosis. Genetic counseling and examination may also be indicated. The purpose of this paper was to present a family case report of nevoid basal cell carcinoma syndrome with multiple odontogenic keratocysts. The features identified by these combined clinical, imaging, and histologic findings are described, along with a brief mention of the family history and a review of the literature. (J Dent Child 2007;74:140-2)

KEYWORDS: ODONTOGENIC KERATOCYST, MULTIPLE, GORLIN-GOLTZ SYNDROME

The odontogenic keratocyst (OKC) is a jaw cyst with an aggressive clinical behavior, including a high recurrence rate.[1,2] The cyst may occur in all ages, but with a peak incidence in the second and third decade of life. Most OKCs present as solitary lesions; however, multiple primary cysts may occur, the most common association being with nevoid basal cell carcinoma syndrome (NBCCS).

The purpose of this paper was to report similar clinical findings in a son and mother with multiple odontogenic keratocysts, where the findings on plain films, computed tomography, and histopathologic examinations are compared and contrasted.

## CASE REPORT

A 13-year-old Chinese male presented for orthodontic treatment to the Department of Orthodontics in School of Stomatology at Shandong University, Jinan, Shandong, People's Republic of China. Panoramic radiographs revealed multiple cysts in the jaws (Figure 1a). The patient was referred to the authors for further evaluation and treatment.

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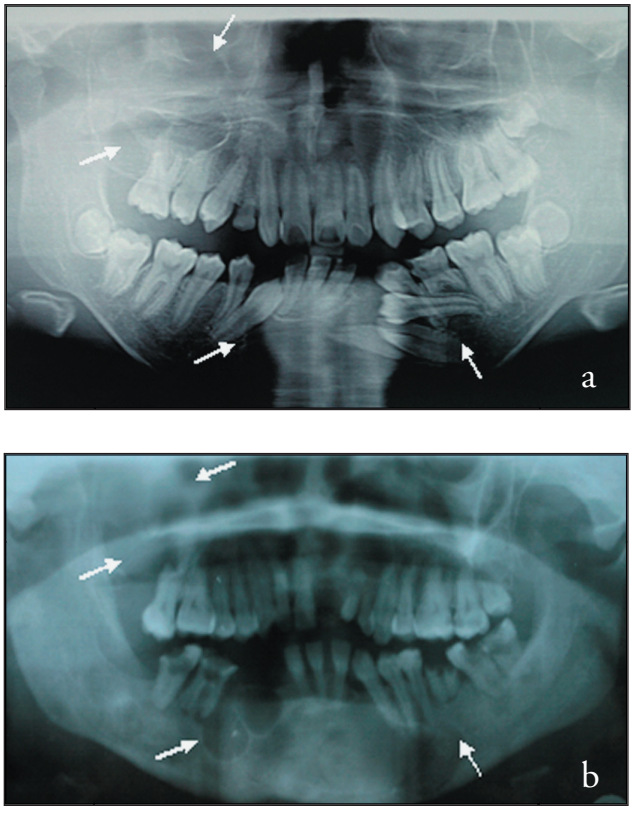


*Figure 1. Panoramic radiograph showed the location and extent of multiple radiolucent lesions: (a) the son; (b) the mother.*

Family history revealed that his mother once underwent an operation to excise multiple cysts in the jaws and associated teeth at the age of 32 years. The panoramic radiograph and computed tomography (CT) scan of the client showed multiple cystic lesion in the maxilla and mandible (Figure 1b). The pathology demonstrated odontogenic and parakeratinized keratocysts. A chromosome study from the peripheral blood showed 46, XX.

An extraoral examination revealed large calvaria, a broadened nasal root, high-arched eyebrows, and mild hypertelorism. An intraoral examination, meanwhile, revealed poor occlusion, malalignment of existing teeth, and 5 unerupted permanent teeth in the right maxilla and the mandibular anterior region.

The panoramic radiograph and CT scan showed multiple cystic lesion in the jaws associated with unerupted teeth. They were on the right maxillary posterior area (Figure 2a) and the mandibular anterior region (Figure 2b). The teeth were impacted to the edge. There was strong ring enhancement. The well-defined border of the right maxillary sinus on the Waters' view of the CT scan suggested a cystic lesion. From these radiographic findings, the authors considered multiple OKCs. These raised a suspicion of NBCCS Syndrome and other relevant investigations were done, but no other associated symptoms were noticed. Chromosomal analysis showed no abnormality.

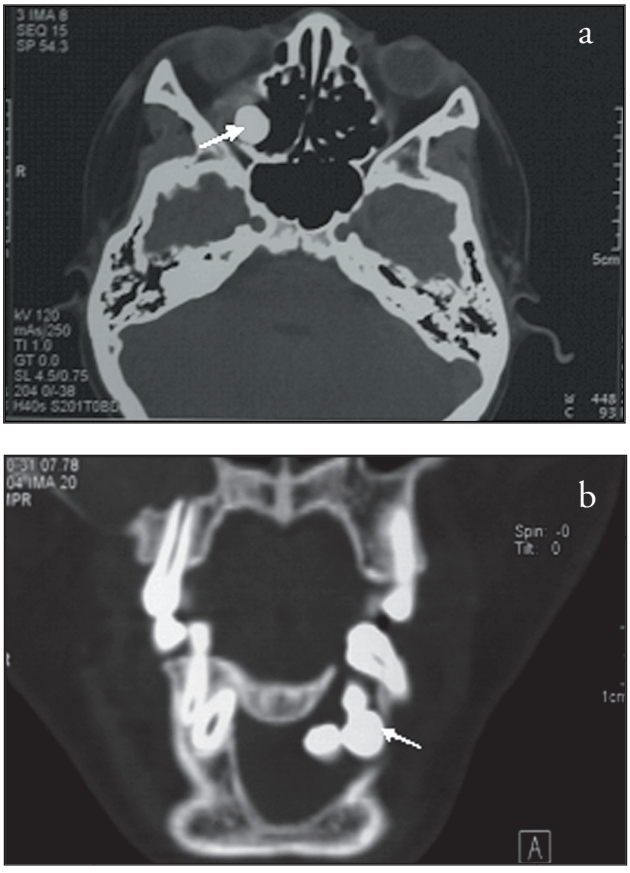


*Figure 2. CT scan of the son showed multiple cystic lesion in the jaws associated with unerupted teeth: (a) the impacted teeth of the right maxillary posterior area; (b) the impacted teeth of the mandibular anterior region.*

Surgical excision with peripheral osseous curettage was planned under a general anesthetic. The cyst appeared to contain a white, cheesy material and contained the crown of a retained tooth within its lumen (Figure 3a). The cyst epithelium proliferated formed a new cyst in the host tissue and retained its typical histological features (Figure 3b). The lesions were histologically reported as keratocysts and parakeratinized.

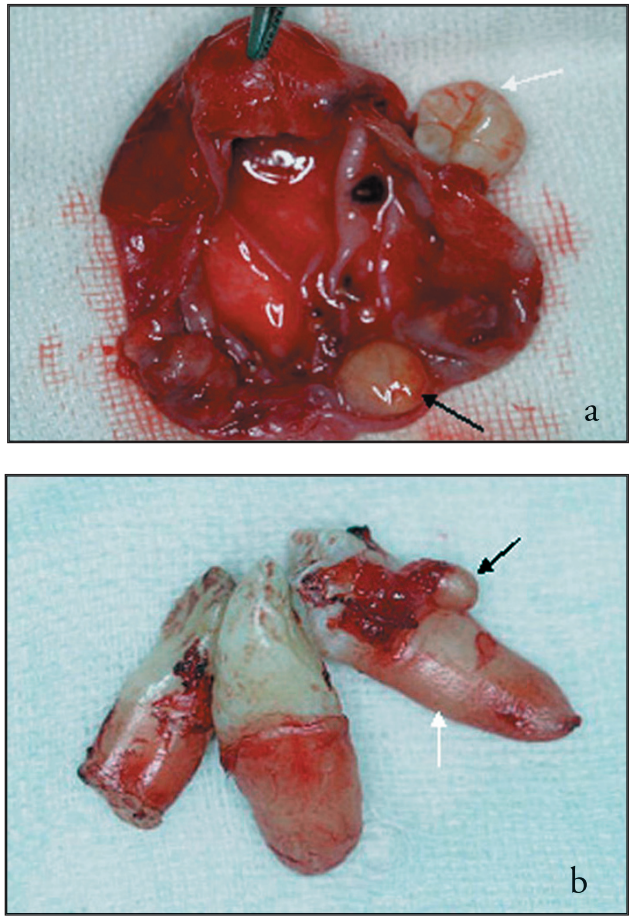


*Figure 3. The impacted teeth of the son after cysts removal: (a) the maxillary cysts were associated with the crown of a tooth and a microcyst; (b) the daughter's microcysts of the mandibular cysts were associated with roots like a lateral periodontal cysts.*

## DISCUSSION

The OKCs are significant clinical entities due to their tendency for recurrence and aggressive behavior.[1,2] Gorlin and Goltz[8] defined this syndrome. The OKC was first described in 1876[3] and named by Phillipsen in 1956.[4] The incidence of OKC among all cysts is reported to be 8%.[5] Some authors suggest that as many as half of OKCs are related to NBCCS.[6] Multiple OKCs are a well-recognized feature of NBCCS.[7] This disorder has an autosomal dominant mode of inheritance. All the clinical manifestations of NBCCS do not have to be present for a diagnosis. In fact, there are authors who think that the presence of multiple keratocysts—even just keratocysts if there is a family association[9]—is sufficient to

establish it. But the majority of authors make the diagnosis if 2 major criteria (OKCs of the jaws, nevoid basal cell carcinomas, palmar and plantar pits, cerebral falx calcifications, family history, and skeletal anomalies) or 1 major and 2 minor criteria (macrocephalus, hypertelorism, medulloblastoma, ovarian calcification) are presented.[10] Early diagnosis and treatment is essential, as well as family detection and genetic counseling, which is important to recognize when a patient has multiple OKCs, because lifelong monitoring is essential for patient management. The authors can anticipate a high risk of recurrence in this study's patient because of the multiple lesions, pathologic findings of parakeratinized keratocysts, and the family history.

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