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Imaging findings in a case of Gorlin-Goltz syndrome: a survey using advanced modalities

Pegah Bronoosh, Ali Reza Shakibafar*, Maneli Houshyar, Shima Nafarzade**

Oral Radiology Department, Dental School, Shiraz University of Medical Sciences, Shiraz, Iran
**TABA Medical Imaging Center, Shiraz, Iran*
***Oral Pathology Department, Babol Dental School, Babol, Iran*

ABSTRACT

Gorlin-Goltz syndrome is an infrequent multi-systemic disease which is characterized by multiple keratocysts in the jaws, calcification of falx cerebri, and basal cell carcinomas. We report a case of Gorlin-Goltz syndrome in a 23-year-old man with emphasis on image findings of keratocyctic odontogenic tumors (KCOTs) on panoramic radiograph, computed tomography, magnetic resonance (MR) imaging, and Ultrasonography (US). In this case, pericoronal lesions were mostly orthokeratinized odontogenic cyst (OOC) concerning the MR and US study, which tended to recur less. The aim of this report was to clarify the characteristic imaging features of the syndrome-related keratocysts that can be used to differentiate KCOT from OOC. Also, our findings suggested that the recurrence rate of KCOTs might be predicted based on their association to teeth. (***Imaging Sci Dent 2011; 41 : 171-5***)

KEY WORDS : Gorlin-Goltz syndrome; Tomography, X-Ray Computed; Magnetic Resonance Imaging; Ultrasonography

Gorlin-Goltz syndrome (nevoid basal cell carcinoma syndrome) is a rare autosomal-dominant disorder and it is associated with various developmental anomalies and the risk of several neoplasms. The most important criteria in diagnosis of this syndrome are the presence of basal cell carcinomas, odontogenic keratocysts, palmar and/or plantar pits, and ectopic calcification of falx cerebri.[1,2] Multiple keratocystic odontogenic tumors (KCOTs), that tend to appear in second decade of life, have been of interest among dentists due to their biological aggressiveness and great amount of recurrence.[3] Recently, these lesions have been reclassified as odontogenic tumors based on the intrinsic growth of epithelial lining.[4]

The radiologic protocol for the diagnosis of this syndrome may include panoramic radiography to detect multiple jaw cysts, skull radiography for evaluation of falx cerebri calcification, chest radiography to detect bifid, fused or splayed ribs, and computed tomography (CT) as well as magnetic resonance (MR) images to find further abnormalities.[2,3] In recent years, the use of sophisticated modalities has been of increasing demand to make a more accurate diagnosis. Exploring the content of lesion, heterogeneity, intra tumoral vascularity, and assessing cortical perforation and soft tissue involvement are possible via advanced imaging techniques.[5]

A survey in the literatures revealed a wide range of signal intensities in MR study of KCOTs and even a little data regarding its sonographic pattern.[6-8] Since it was valuable to determine whether there are radiographic features that could predict the risk of recurrence, we evaluated various imaging modalities, in a case of multiple KCOT and compared it with the previously published data.

Case Report

A 23-year-old man was referred to our department to take radiographs for orthodontic treatment. Accidentally, multiple radiolucent lesions were found on panoramic radiograph (Fig. 1) and there was an evidence of bone defect in the premaxilla. Postero-anterior skull radiograph showed falx cerebri calcification (Fig. 2).

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Correspondence to : Dr. Pegah Bronoosh
Oral Radiology Department, Shiraz Dental School, Qasrodasht Street, Shiraz 7195615878, Iran
Tel) 98-711-6263192, Fax) 98-711-6270325, E-mail) pegah_brix@yahoo.com

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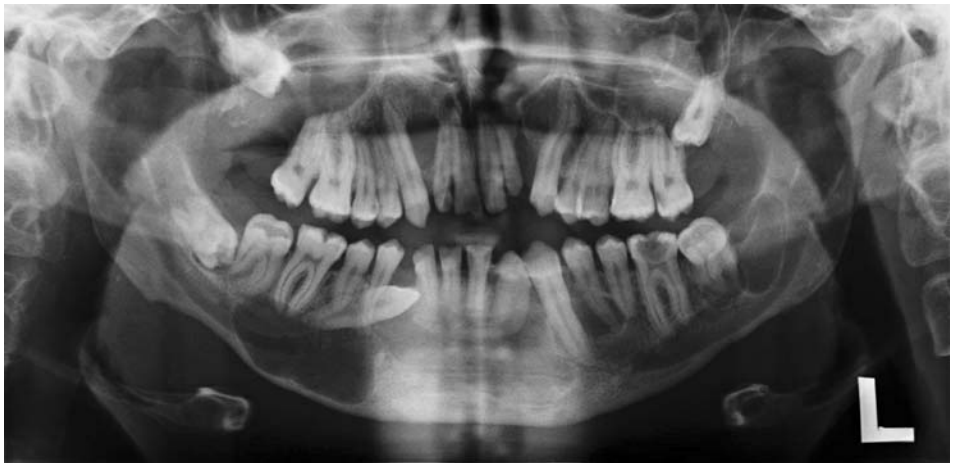

Fig. 1. Panoramic radiograph shows multiple cystic odontogenic lesions in both jaws.

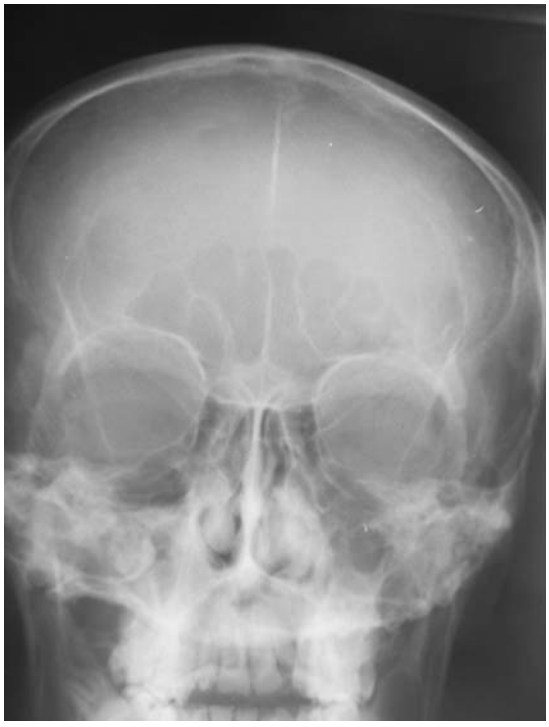

Fig. 2. Skull PA radiograph shows falx cerebri calcification.

Clinical examination revealed a frontal bossing and hypertelorism as well as an evidence of previous lip closure. No skin lesion was detected. Two major criteria (multiple jaw cysts and falx cerebri calcification) and some congenital anomalies (cleft lip and palate, frontal bossing, and moderate hypertelorism) strengthened the possibility of Gorlin-Goltz syndrome. Patient referred for advanced imaging evaluation.

CT scan showed an expansile lytic lesion in the right antrum pericoronal to the impacted tooth (3×3 cm) and a small lytic lesion between maxillary lateral incisor and canine (Figs. 3A and B). There was a mucous retention pseudocyst in the left antrum with iso-attenuation (Fig. 3C). Three isolated expansile lytic lesions were found in the mandible (Fig. 3D). Expansion and thinning of the cortex was found on the medial wall of the left ramus (Fig. 3E).

Four out of five lesions were detected using ultrasonography (US) (Fig. 4). All lesions were well defined hypoechoic in B mode except the right maxillary cyst with a mixed echogenicity. None of the lesions had signal in Doppler mode. One lesion was not detected due to the thick overlying bone.

MR study was performed on the axial and coronal T1, T2 weighted images (WI), and pre- and post-contrast T1WI. Three isolated lesions were detected in the mandible. Lesions showed low to intermediate signal intensity on T1WI with moderate peripheral enhancement (Figs. 5A-C). On T2WI, the lesions had high signal intensity with internal homogeneity (Fig. 5D). There was an expansile, well defined, and hyper intense lesion in the right maxillary sinus with heterogeneous signal on T2WI due to high protein content. Incidentally, a mucous retention pseudocyst in the left maxillary sinus was found with a homogenous intermediate signal on T1WI without peripheral enhancement and high signal intensity on T2WI (Figs. 5E and F).

On histopathologic examination, 4 lesions reported to be of parakeratinized KCOT and the lesion in the right maxillary sinus was orthokeratinized odontogenic lesion (Fig. 6).

Discussion

KCOT represents from 3 to 15% of all odontogenic lesions and appears in 65-75% of cases of the syndrome. This lesion has been of interest due to its biological aggressiveness and high reported rate of recurrence.[9] On radiographs,

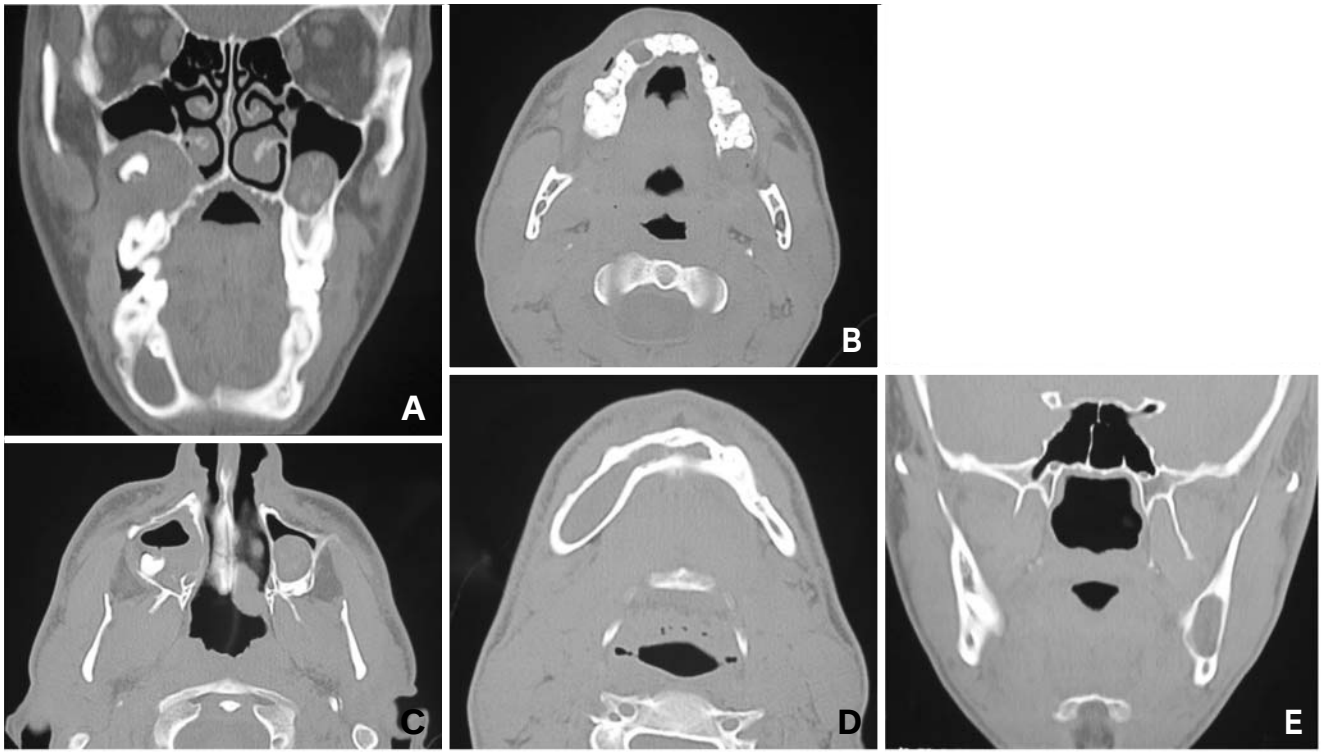

Fig. 3. A. Coronal CT scan shows a cystic lesion associated with an impact tooth causing expansion and displacement of the floor of the right antrum and a mucous retention cyst in the left sinus. B. Axial CT scan shows a lytic lesion in the region of right maxillary canine as well as in ramus both sides. C. Axial CT scan shows soft tissue density in the right maxillary sinus with mucoperiosteal thickening and thinning of medial wall. D. Axial CT scan of the mandible reveals a well defined expansile lytic lesion in the right side crossing midline. E. Coronal CT scan shows a hypodense expansile lesion causing thinning of medial wall of the left mandibular ramus.

KCOTs appear as uni/multilocular radiolucent lesions that may have a smooth or scalloped border.[3,6] Cortical expansion and thinning and rarely a hazy lumen have been reported in the previous reports.[6] The lesions sometimes occur around an impacted tooth.[4]

Nevertheless, few studies have assessed ultrasonographic appearances of KCOTs. Lauria et al[10] reported that KOTs were hypoechogenic in 7 of 9 cases because of their dense and thick content, and 2 other lesions showed solid content in their study on ultrasonographic evaluation of bone lesions of the jaw. In 2010, Bagewadi et al[11] stated a homogenous hypoechoic internal echo pattern for this lesion. In 2009, Sumer et al[12] evaluated 4 odontogenic keratocysts by ultrasonography, and for the first time, by Doppler mode. Three lesions had complex, dense echo without internal vascularization which was consistent with the findings by Lauria et al. The other lesion was not detected due to the thick vestibular bone. In the present study, we also used Doppler mode to check internal vascularization. Three lesions were hypoechoic without Doppler signal similar to the previous studies. The lesion in the right maxillary sinus which was associated with an impacted tooth had a mixed pattern in US study.

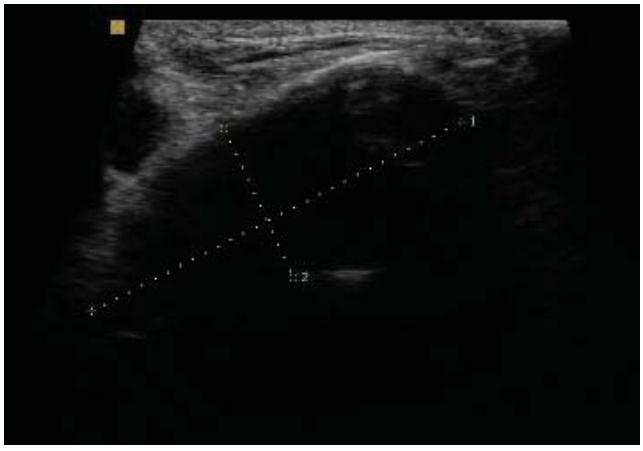

Fig. 4. Ultrasonograph shows a well demarcated hypoechoic lesions in the right side of mandible.

MRI of KCOT shows various signal intensities. In 2002, Hisatomi et al[7] reported an intermediate-to-high signal intensity on T1WI (T1 weighted image) and heterogeneous low-high signal intensity on T2WI and stated that these signal intensities on MRI might be suggestive of KCOT.

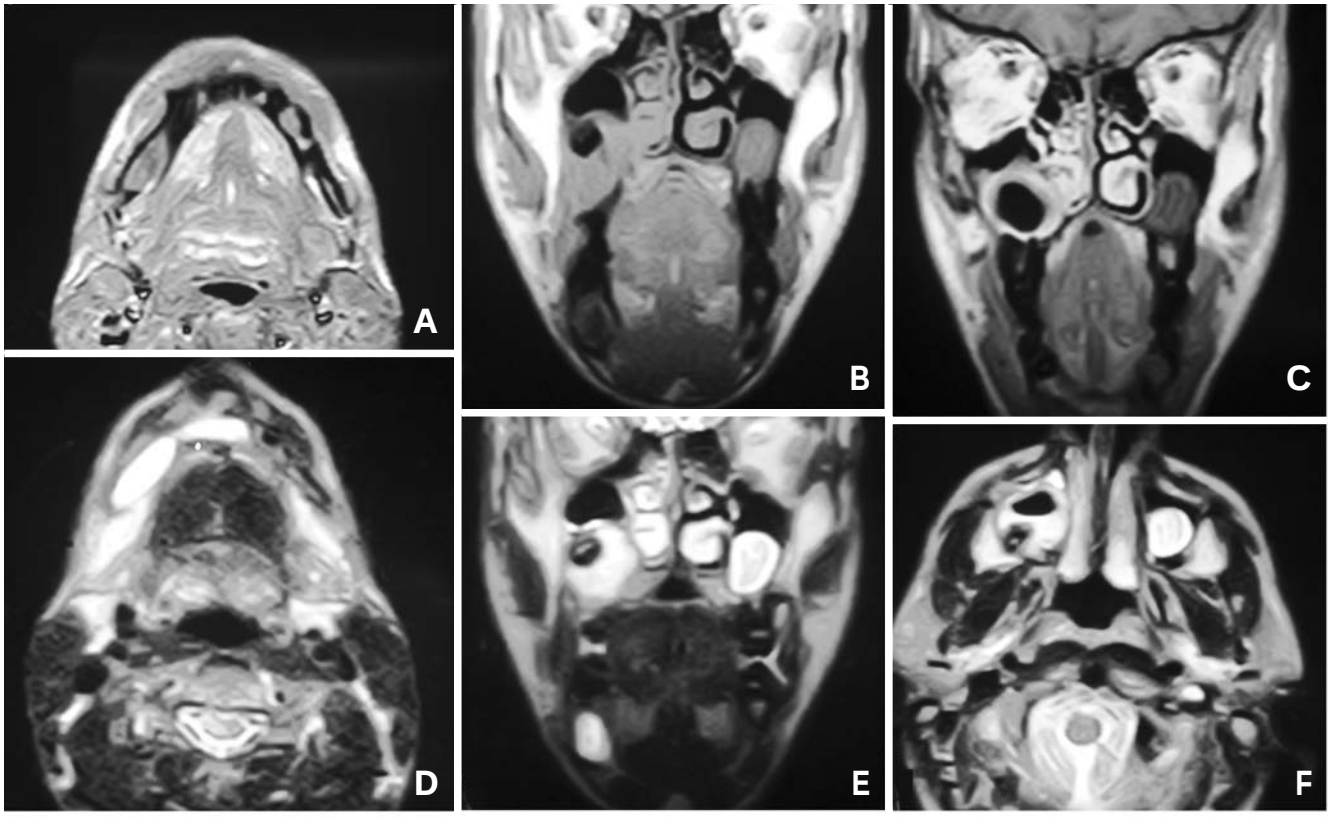

Fig. 5. A. Axial T1WI of mandible shows intermediate signal intensity (SI). B. Coronal T1W MRI of patient shows an expansion of inferior wall of the right maxillary sinus and anterior wall of nasal cavity due to low-intermediate SI lesion associated with an impacted tooth (signal void). C. Coronal post contrast T1W MRI reveals moderate enhancement of cystic wall in the right maxillary sinus. D. Axial T2WI of the mandible shows a homogenous high SI lesion. E. Coronal T2W MRI reveals a well defined expansile hyperintense lesion inferior to the right antrum with heterogeneous signal intensity. F. Axial T2WI of the maxilla shows a hyper intense heterogeneous lesion and a retention pseudocyst.

Minami et al[6] in 1996 showed that odontogenic keratocysts had intermediate or high signal intensity on T1WI and intermediate signal intensity on T2WI. In the present case, all lesions had high signal intensity on T2WI except for the right maxillary lesion that showed heterogeneous high signal intensity on T2WI. Sometimes the lumen of KCOT contains a large quantity of keratin. If the lesion was secondarily infected, cholesterol and hyaline bodies might be found. Therefore, the signal intensity on T1/T2WI might reflect these contents.

In 2005, the World Health Organization (WHO) classified the parakeratinized odontogenic tumor as KCOT and stated that orthokeratinized cystic lesions (OOC) were separate entity.[13,14]

KCOT is the lesion that occurs in Gorlin's syndrome.[13,15,16] Bolbaran reported a case of orthokeratized type in basal cell nevoid syndrome,[15] however KCOTs associated with syndromes often were of parakeratinized type and had a much higher recurrence rate compared with the isolated ones.[2,13] McDonald-Jankowski reported that orthokeratinized odontogenic lesions occurred with an unerupted tooth

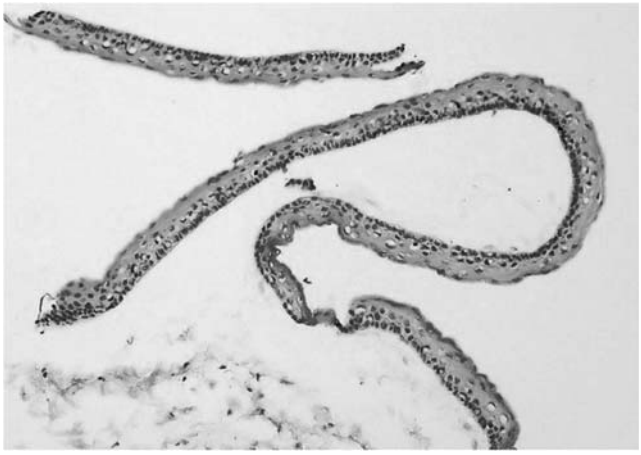

Fig. 6. Histopathologic view of KCOT (H&E stain, ×10). The epithelial lining is 6 to 8 cells thick, with a hyperchromatic and palisaded basal cell layer. Note the corrugated parakeratotic surface.

significantly more than parakeratinized ones.[13,17] This may indicate a different pathogenesis for lesions associated with impacted teeth and probably a different keratin content which is reflected in MR and US studies.

In this case, the pericoronal lesion in the right maxillary sinus revealed the different echo pattern compared with others, reported to be an OOC. It seems that pericoronal lesions may have different internal keratin density which is responsible for its heterogenous appearance in MR and mixed echo pattern in US studies. Unfortunately, the previous studies did not indicate whether KCOTs were related to the impacted teeth or not.

Crowley et al stated the recurrence rates of KCOTs (42.6 %) and OOCs (2.2%).[18] Therefore, it is also important from the treatment aspect, since the parakeratinized lesions had more aggressive behavior to decrease the recurrence rate.[13] However, the rate of recurrence in syndrome associated lesions is higher than the isolated ones.[9]

In conclusion, KCOTs might represent various imaging features, especially using sophisticated modalities such as MR and US studies. These features might predict the aggressiveness of KCOTs which was the most helpful planning a surgical and follow up protocol. In addition, our US findings were compatible with the findings in CT and MR studies. Therefore, US study provides to evaluate the size and content of maxillofacial lesions. It is important to establish the diagnosis as soon as possible in syndrome-related KCOTs due to the susceptibility of these patients to fatal neoplasms.

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