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CASE REPORT

Multiple Keratocysts of the Mandible in Association with Multiple Basal Epithelioma, Jaw Cysts and Bifid Rib Syndrome: A Rare Case Report

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ABSTRACT

Nevoid basal cell carcinoma syndrome is a syndrome with wide variety of manifestations ranging from oral lesions to skeletal deformities. It calls for due responsibility of maxillofacial surgeon to diagnose the syndrome because very often they are the first health professionals to see the patient for the treatment of keratocystic odontogenic tumor.

Keratocystic odontogenic tumor (KCOT) has been the topic of numerous investigators and is known for its potentially aggressive behavior and significant rate of recurrences. Keratocystic odontogenic tumor often occurs as a solitary lesion, and in some instances multiple keratocysts may occur in association with a syndrome called Gorlin-Goltz syndrome (nevoid basal cell carcinoma, jaw cyst bifid rib basal cell nevus syndrome).

Here, we present a case of multiple keratocysts in the mandible in association with skeletal, ocular and cutaneous anomalies in the given clinical scenario which has profound relevance in the clinical dental practice.

Keywords: Syndromic multiple keratocysts, Mandible, Nevi, Gorlin-Goltz syndrome.

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INTRODUCTION

The nevoid basal cell carcinoma syndrome, first delineated by Gorlin and Goltz (1960) is characterized by basal cell carcinoma, odontogenic keratocysts, palmar and plantar pits and ectopic calcification of falx cerebri.[1,2] Seventy-five percent of patients affected by nevoid basal cell carcinoma syndrome (NBCCS) show multiple and bilateral keratocysts. Mainly located in premolar area may displace teeth with consequent malocclusion.[2]

This syndrome has received several names throughout the times such as 'basal cell nevus syndrome', 'nevoid basal cell carcinomas syndrome' or the most complex name of 'multiple basal epithelioma, jaw cysts and bifid rib syndrome'.[3]

Nevoid basal cell carcinoma syndrome is a rare autosomal dominant condition that can cause unusual facial appearances. The reported prevalence is one case per 56000-164000.[1,4] Despite the number of cases reported in the literature, the understanding of complete form of NBCCS is not yet conclusive. Besides the fact that the signs and symptoms of NBCCS appear as the patient ages, they do not occur concomitantly; these are challenges to the diagnostician.[5]

The dental clinician may be the first to encounter and identify this syndrome when multiple cysts like radiolucencies are discovered on radiographic examination of the jaws. Due to the importance of oral and maxillofacial manifestations of this syndrome, it is fundamental to know its characteristic in order to make a diagnosis, an early preventive treatment and establish right genetic advice.[6] This piece of work attempts to highlight the salient features of an unusual case of multiple keratocysts in association with Gorlin-Goltz syndrome with its management.

CASE REPORT

In August 2012, a 21 years old male reported to the OPD, Department of Oral and Maxillofacial Surgery, Tatyasaheb Kore Dental College and Research Centre, Kolhapur, Maharashtra, India, with chief complaint of pain and swelling in the lower left back region of the jaw since 3 to 4 months. The patient had visited a dentist few months ago with the same complaint where removal of 36, 37 was advised. Patient was moderately built and well nourished with no history of chewing habits and nonsignificant medical and dental history.

General physical examination revealed bluish black macular pigmentation measuring 0.25 × 0.75 cm on forehead

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and periorbital region was evident (Fig. 1). Clinical examination revealed mild facial asymmetry on the left side of face. Intraoral examination showed significant expansion of the buccal cortex, egg shell crackling, overlying swelling was firm in consistency, tender, nonfluctuant and nonpulsative upon palpation.

The patient was subjected for radiographic examination which revealed well defined radiolucent areas extending on either sides of mandibular body, angle and ramus region respectively. Significant resorption of the bone was evident on left side in relation to 35, 36, 37 region respectively (Fig. 2).

Observing the OPG findings a radiographic diagnosis of multiple keratocystic odontogenic tumors was suggested.

The patient was subjected for fine-needle aspiration cytology (FNAC) and incisional biopsy following the routine blood examination which helped us to arrive to a final diagnosis keratocystic odontogenic tumor in association with Gorlin-Goltz syndrome (Fig. 3). Patient was taken up for surgical enucleation of the cystic lesion on both sides with application of Carnoy's solution onto the defect under general anesthesia (Figs 4A and B). Patient was periodically followed up for the recurrence of disease and was monitored for regeneration of the bone defect accordingly.

Fig. 1: Skin lesions on the forehead and periorbital region

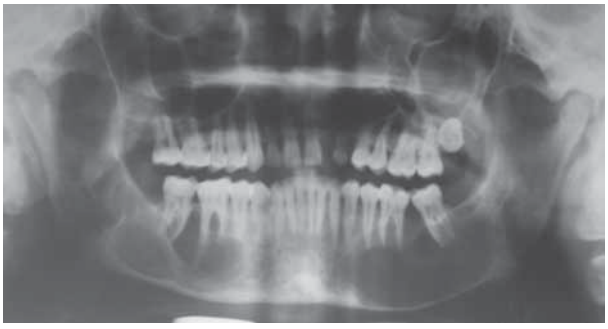


Fig. 2: Panoramic radiograph showing multilocular appearance of the lesion

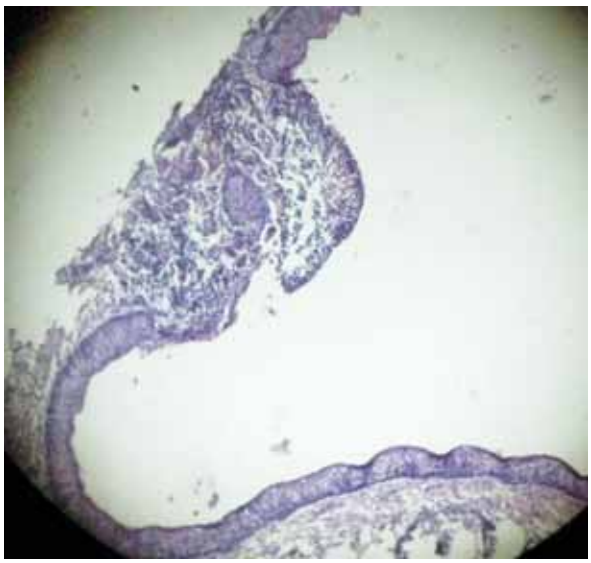


Fig. 3: Photomicrograph showing characteristic lining of keratocystic odontogenic tumor (H&E stain)

DISCUSSION

Nevoid basal cell carcinoma syndrome is a rare inherited multisystem disorder that is a result of mutations in the PTCH gene. More than 100 clinical abnormalities have been reported in this syndrome. The major criteria are—early development of multiple basal cell cancers (BCCs), odontogenic (bone) keratocysts, palmar and plantar pitting, ectopic intracranial calcification and family history. Minor criteria include craniofacial anomalies (macrocephaly, frontal bossing, hypertelorism), bifid ribs, early onset medulloblastomas, cardiac or ovarian fibromas, lymphomesenteric cysts and congenital malformations (cleft lip/palate, polydactyly, eye abnormalities, colobomas, cataracts and glaucoma).[7-9]

Major criteria are as follows:

- More than two BCCs or one under age of 20 years
- Odontogenic keratocyst
- Three or more palmar pits
- Bilamellar calcification of falx cerebri
- Bifid, fused or splayed ribs
- First-degree relative with NBCCS

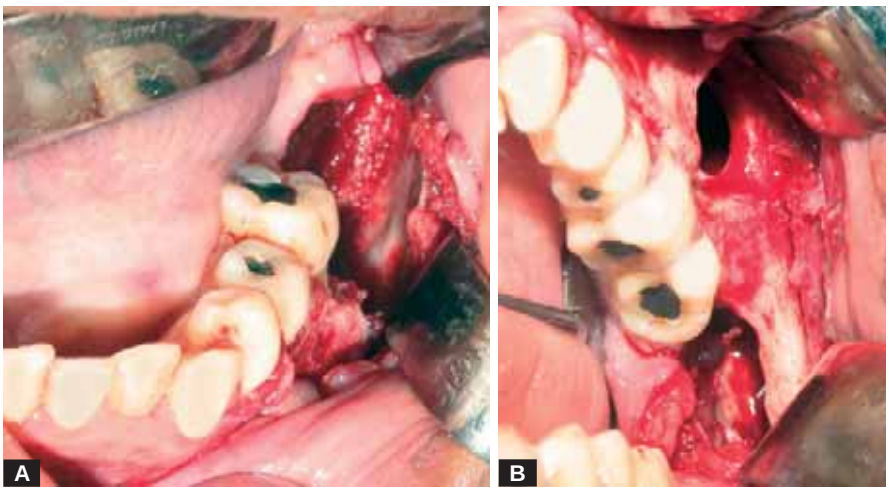
Minor criteria are as follows:

Macrocephaly adjusted for height:

- Frontal bossing, cleft lip/palate and hypertelorism
- Sprengel deformity, pectus and syndactyly of digits
- Bridging of sella turcica, hemivertebrae and flame shaped
- Radiolucencies
- Ovarian fibroma
- Medulloblastoma

The diagnosis is supported by finding either two major, or one major and two minor criteria.[9]

The odontogenic keratocyst is an epithelial developmental cyst of the jaw.[10] Some surgeons believe the cyst can be properly treated with enucleation if the lesion is



Figs 4A and B: Intraoperative views of the surgical site

removed intact.[11,12] However, complete removal of the OKC can be difficult because of the thin friable epithelial lining, limited surgical access, skill and experience of the surgeon, cortical perforation, and the desire to preserve adjacent vital structures.[13]

Less than 10% of patients with multiple keratocysts have other manifestations of this syndrome; however, it has been suggested that multiple keratocysts alone may be the confirmatory of this syndrome. In the present case, surgical enucleation along with chemical cauterization was done which holds well with the statement that chemical cauterization is a proved adjunctive technique in case of keratocysts and is useful to prevent recurrence by fixing the daughter cysts or remnants of epithelial lining during the enucleation procedure.[14]

CONCLUSION

It can be said that Gorlin-Goltz syndrome associated with multiple keratocysts is of particular interest for the oral maxillofacial health experts. Thorough clinical examination supplemented with appropriate investigations would reveal the concerned diagnosis as evidenced in the present case. Early diagnosis will often make it possible to use conservative therapies rather than complex treatments. Furthermore, it offers patients and their families the chance of discovering the possible hereditary risks of the condition.

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