Multiple keratocysts of the mandible in association with Gorlin-Goltz 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 has been the topic of numerous investigators, is known for its potentially aggressive behavior, significant rate of recurrences. KCOT often occurs as a solitary lesion, in some instances multiple keratocysts may occur in association with a syndrome called Gorlin-Goltz syndrome (nevoid BCC, jaw cyst bifid rib basal cell nevus syndrome). Here, we present a case of multiple keratocysts in the mandible in association with skeletal, ocular, cutaneous anomalies in the given clinical scenario, which has profound relevance in the clinical dental practice.

Keywords: Gorlin-Goltz syndrome, mandible, naevi, syndromic multiple keratocysts

Introduction

The nevoid basal cell carcinoma syndrome, first delineated by Gorlin Goltz (1960) is characterized by BCC, odontogenic keratocysts (OKC), palmar, plantar pits, ectopic calcification of falx cerebri.[1,2] 75% of patients affected by NBCCS show multiple 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," "NBCCS," or the most complex name of "multiple basal epithelioma, jaw cysts bifid rib syndrome."[3]

Nevoid basal cell carcinoma syndrome is a rare autosomal dominant condition that can cause unusual facial appearances.

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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 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 identify this syndrome when multiple cysts like radiolucencies are discovered on radiographic examination of the jaws. Due to the importance of oral maxillofacial manifestations of this syndrome, it is fundamental to know its characteristic in order to make a diagnosis, an early preventive treatment 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-year-old male reported to the out-patient Department of Oral Maxillofacial Surgery, Tatyasaheb Kore Dental College Research Centre, Kolhapur, Maharashtra, India with the chief complaint of pain swelling in the lower left back region of the jaw since 3-4 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 well-nourished with no history of chewing habits, non significant medical dental history.

General physical examination revealed bluish black macular pigmentation measuring 0.25 × 0.75 cm on forehead, periorbital region was evident [Figure 1]. Clinical examination revealed mild facial asymmetry on the left side of face. Intra oral examination showed significant expansion of the buccal cortex, egg shell crackling; Overlying swelling was

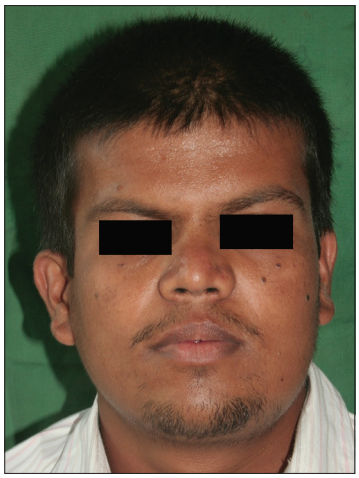

Figure 1: Skin lesions on the forehead and peri orbital region

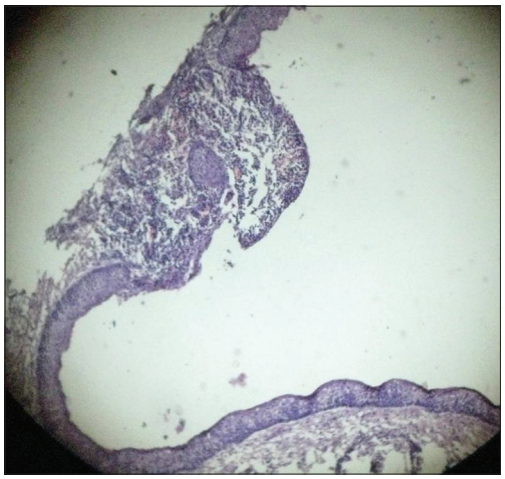

Figure 3: Photomicrograph showing charcteristic lining of keratocystic odontogenic tumor (H and E)

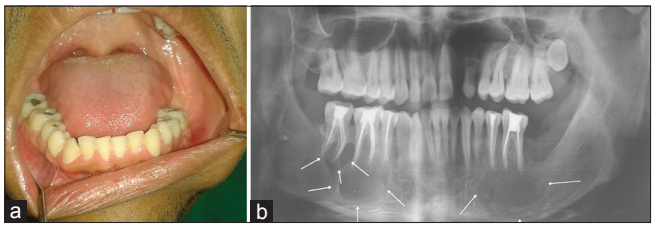

Figure 5: (a) Postoperative clinical view of the operated site showing satisfactory wound healing. (b) Postoperative radiographic view of the site showing satisfactory bone regeneraion

firm in consistency, tender, nonfluctuant, nonpulsative upon palpation.

Further the subjected for radiographic examination which revealed well-defined radiolucent areas extending on either sides of mandibular body, angle, ramus region, respectively. Significant resorption of the bone was evident on left side in relation to 35, 36, 37 region respectively [Figure 2].

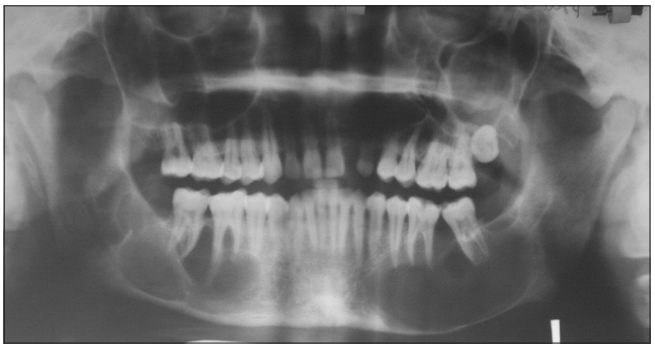

Figure 2: Panoramic radiograph showing multilocular appearance of the lesion

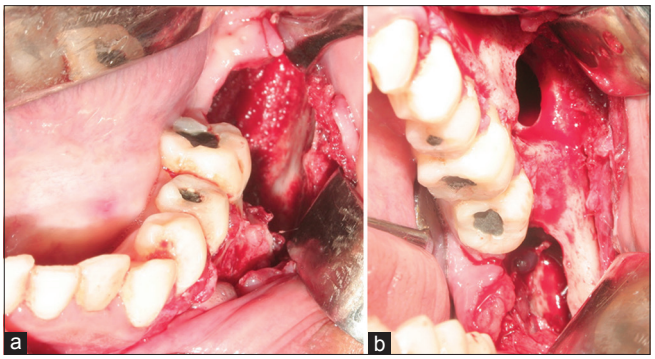

Figure 4: (a and b) Intraoperative view of the surgical site

Observing the orthopantomograph findings a radiographic diagnosis of multiple keratocystic odontogenic tumors (KCOTs) was suggested.

The patient was subjected for fine-needle aspiration cytology, incisional biopsy following the routine blood examination, which helped us to arrive to a final diagnosis KCOT in association with Gorlin-Goltz syndrome [Figure 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 [Figure 4a and b]. Patient was periodically followed-up for the recurrence of disease was monitored for regeneration of the bone defect accordingly [Figure 5a and b].

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 BCCs; odontogenic (bone) keratocysts; palmar and plantar pitting; ectopic intracranial calcification; family history. Minor criteria include: Craniofacial anomalies (macrocephaly, frontal bossing, and hypertelorism); bifid ribs; early onset medulloblastomas; cardiac or ovarian fibromas; lymphomesenteric cysts; congenital malformations (cleft lip/palate, polydactyly, eye abnormalities, colobomas, cataracts, and glaucoma).[7-9]

Major criteria are as follows:
- More than 2 BCCs or one under age of 20 years
- OKC
- 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 flameshaped
- Radiolucencies
- Ovarian fibroma
- Medulloblastoma.

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

The OKC is an epithelial developmental cyst of the jaw.[10] Some surgeons believe the cyst can be properly treated with enucleation if the lesion is 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 this 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 is useful to prevent recurrence by fixing the daughter cysts or remnants of epithelial lining during the enucleation procedure.[14]

Conclusion

As a 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 their families the chance of discovering the possible hereditary risks of the condition.

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